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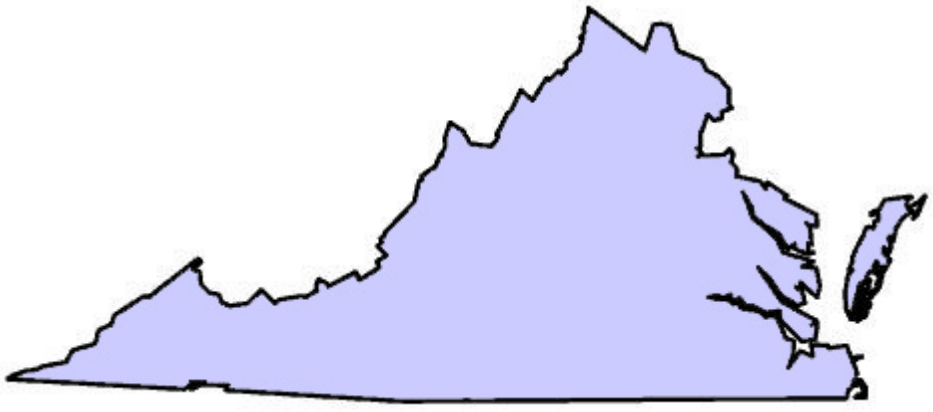
Virginia GeneSEAN

# Genetic Services and Education – An Assessment of Needs

*A Comprehensive Assessment of Medical Genetic Services and Education in the Commonwealth of Virginia*



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## LIST OF ACRONYMS

AAFP ----------American Academy of Family Physicians
AAP----------American Academy of Pediatrics
ABGC ----------American Board of Genetic Counseling
ABMG----------American Board of Medical Genetics
ABMS ----------American Board of Medical Specialties
ACMG----------American College of Medical Genetics
ACOG ----------American College of Obstetrics and Gynecology
ADA ----------Americans with Disabilities Act
AMA----------American Medical Association
ANA ----------American Nurses Association
APHMG----------Association of Professors of Human and Medical Genetics
ASCO----------American Society of Clinical Oncology
ASHG----------American Society of Human Genetics
ASTHO----------Association of State and Territorial Health Officials
CAH ----------Congenital adrenal hyperplasia
CARN ----------Collaborative Ambulatory Research Network
CATPIP ----------Congenital Anomalies Tracking and Prevention Improvement Project
CCC----------Care Connection for Children
CDC----------Centers for Disease Control and Prevention
CEGEN----------Confidential Enquiry into Counseling for Genetic Disorders
CHKD ----------Children's Hospital of The King's Daughters
CORN ----------Council of Regional Networks for Genetic Services
CSHCN----------Children with Special Health Care Needs
CSS ----------Children's Specialty Services
DNA ----------Deoxyribonucleic acid
EVMS ----------Eastern Virginia Medical School
FAMIS----------Family Access to Medical Insurance Security
FH ----------Familial hypercholesterolemia
FOPE II----------Future of Pediatric Education II
GEC----------Genetics Education Center
GENA ----------Genetic Education for Native Americans
GeneSEAN----------Genetic Services and Education - an Assessment of Needs
GEP ----------Genetic Education Project
GIFT ----------Genetics Interdisciplinary Faculty Training
GIVF----------Genetics and IVF
GPC ----------Genetics in Primary Care
GROW----------Genetic Resources on the Web
HGP----------Human Genome Project
HIPAA----------Health Insurance Portability and Accountability Act of 1996
HMO----------Health Maintenance Organization
HNPCC----------Hereditary non-polyposis colon cancer
HRSA----------Health Resources and Services Administration
HSA----------Health Services Area
IPA ----------Independent Practice Association
IRB----------Institutional Review Board
ISONG----------International Society of Nurses in Genetics

IVF ----------In Vitro Fertilization
LEGENDS ----------Learning Exchange for Genetic and Environmental Disease Solutions
LEND----------Leadership Education in Neurodevelopmental and related Disabilities
MCAD----------Medium-chain acyl-CoA dehydrogenase deficiency
MCHB ----------Maternal and Child Health Bureau
MCV ----------Medical College of Virginia
OD ----------March of Dimes
MR ----------Mental retardation
NCHPEG----------National Coalition for Health Professional Education in Genetics
NHGRI ----------National Human Genome Research Institute
NICU ----------Neonatal Intensive Care Unit
NMCP ----------Naval Medical Center Portsmouth
NNSGRC----------National Newborn Screening and Genetic Resource Center
NSGC----------National Society of Genetic Counselors
OMIM ----------Online Mendelian Inheritance in Man
ORD ----------Office of Rare Diseases
PCP ----------Primary care provider
PGD----------Preimplantation genetic diagnosis
PKU----------Phenylketonuria
POS ----------Point of Service Plans
PPO ----------Preferred Provider Organizations
PSGS----------Pediatric Screening and Genetic Services
QA ----------Quality assurance
RNA ----------Ribonucleic acid
RPC ----------Regional Perinatal Councils
SIDS ----------Sudden Infant Death Syndrome
SOL ----------Standards of Learning
TFAH ----------Trust for America's Health
TIPS----------Tracking Infant Progress
U.K. ----------United Kingdom
U.S. ----------United States
UVA ----------University of Virginia
VaCARES----------Virginia Congenital Anomalies Reporting and Education System
VaGAC ----------Virginia Genetic Advisory Committee
VASCAP ----------Virginia Sickle Cell Awareness Program
VCU ----------Virginia Commonwealth University
VDH ----------Virginia Department of Health
VISITS ----------Virginia Infant Screening and Infant Tracking System
WHO----------World Health Organization
WiSSP ----------Wisconsin Stillbirth Service Program

# EXECUTIVE SUMMARY

Genetics is playing an ever-increasing role in the practice of healthcare. Particularly with the insights gained from the Human Genome Project (HGP), this role is likely to grow further. The development of new technologies and clinical applications will test existing systems of genetic healthcare. To ensure that Virginia has a strong infrastructure that is equipped to meet the growing challenges of genetic medicine, state leaders have initiated the development of a state genetic plan. Since thoughtful, comprehensive planning requires an understanding of current systems and needs, a group of genetic professionals at Virginia Commonwealth University (VCU) initiated the *Virginia GeneSEAN: Genetic Services and Education - An Assessment of Needs* project in 2003 with grant funding from the Virginia Chapter of the March of Dimes (MOD). The needs assessment was completed in 2005. The main goals, objectives, methodology, and key findings of the project, detailed more extensively throughout this report, are summarized below.

## Study Purpose and Goals

The ultimate goal of the Virginia GeneSEAN project is to assure that high quality systems of care and interventions are available so that all Virginians can access appropriate services for genetic health concerns. Under this ideal, the purpose of the project is to provide a comprehensive assessment of medical genetic services and education throughout Virginia, specifically addressing existing systems of care, prevention services, educational activities, insurance issues, data collection, and quality assurance (QA), as well as gaps in these systems. The results of Virginia GeneSEAN are intended to be used as a framework for the development of an informed state genetic plan. Healthcare professionals, policymakers, advocates, and other stakeholders are encouraged to use the information contained in this report to make health promotion efforts more effective, to help more people gain access to quality genetic services, and to ensure that personal genetic information is protected.

## Study Objectives and Methodology

To accomplish the goals of Virginia GeneSEAN, the project team focused on the following objectives:

- Describe the current status of genetic services and education in the state as well as barriers to high quality and appropriate community-based, family-centered genetic care and information by reviewing existing data sources.
- Assess the current and emerging practices and gaps within genetic health services and education delivery in Virginia through consumer focus groups and surveys of genetics professionals, primary care providers (PCP), insurers, Virginia Department of Health (VDH) staff, and other stakeholders.
- Obtain the participants' input on ways to improve the existing system and address the current and growing disparities in access to genetic services, education, and testing.

Multiple activities have been undertaken over the past three years to meet these objectives. During the first year of the project, a comprehensive review of the medical literature and existing data sources relevant to genetic services and education in Virginia, including a detailed assessment of insurance coverage for genetic services through an examination of insurance organization publications, was performed. Simultaneously, a detailed survey was developed and distributed to all identified clinical genetic providers throughout Virginia. The survey addressed practice patterns and service provision, as well as providers' perceptions of a variety of factors, including the quality and accessibility of genetic services in Virginia, availability of resources, reimbursement issues, legal and ethical issues, the role of

state organizations, and educational needs and practices. In 2004, a brief survey assessing PCPs' perceptions of genetic services and education was developed and distributed to PCPs and health department district directors throughout Virginia. An assessment of genetic consumers' views regarding the quality and accessibility of genetic services in Virginia, genetic services needs, educational needs and practices, and genetic discrimination and privacy was also undertaken in 2004 through focus groups conducted in two regions of the state. The findings of these activities were then analyzed and are summarized in this report. The Council of Regional Networks for Genetic Services (CORN) has developed a framework for conducting needs assessments, and those guidelines are used to illustrate gaps and highlight successes in Virginia's systems of care, while insights gained from an analysis of other states' genetic needs assessment reports, genetic plans, and resulting initiatives are included in this report as well. In the final year of this project, input was gathered from the citizens of Virginia through the development of a Commonwealth Poll, a telephone survey of randomly-selected adults, to identify the general public's knowledge of genetics, their usage of genetic services, issues regarding the location of genetic centers, and their perception of the importance of genetic services. Finally, a brief survey was developed and distributed to individuals representing identified groups of stakeholders that included representatives of insurance companies, specialty doctors (i.e. oncologists, surgeons, cardiologists), allied healthcare professionals (i.e. nurses, nurse practitioners), and major employers in Virginia; representatives of regional hospitals, medical schools, and organizations; government representatives; and representatives from the education system and the media. The survey aimed to gather information on their knowledge of genetics, their opinion on the average Virginian's knowledge of genetics, and the current status of genetic services.

## Key Findings

The key insights obtained are summarized below.

- Genetic factors contribute to almost all human disease.
  - 3-5% of all births are complicated by birth defects.
  - Birth defects are the leading cause of infant deaths.
  - 71% of pediatric hospital admissions are related to conditions that have a genetic basis.
  - Most common adult diseases are at least partially caused by inherited susceptibilities.

- Existing genetic services in Virginia are generally of high quality.
  - Virginia has comprehensive genetic centers mainly located in the largest metropolitan areas, while some areas of the state are served by outreach clinics.
  - Genetic population screening programs operated by VDH, such as newborn screening services and the birth defects registry, have generally received good ratings on national comparisons.
  - Most genetic providers surveyed indicated that existing genetic services in Virginia--including the quality of the genetic workforce, newborn screening services and other population screening systems, and existing genetic outreach services--are examples of successful genetic services in the state.
  - The majority of participants in the genetic consumer focus groups and PCP survey reported being satisfied with existing genetic services in the state, and believe the quality of services is above average.

- The availability of genetic services to the citizens of Virginia is important.
  - The general public overwhelmingly expressed the value of having genetic services available.
  - The majority of key stakeholders viewed genetic services as being vital and expressed exploring

telemedicine capabilities and increasing the number of genetic providers as ways to improve availability.
  - Ten percent of the general public reported using genetic services in the past five years.

- Significant barriers increasingly challenge the availability of family-centered, culturally competent genetic care in Virginia.
  - Unmet genetic services needs remain throughout the state and are particularly acute in Northern Virginia and rural locations, where patients must wait as long as five months for genetic services.
  - Near half of Virginia citizens will only pursue genetic services if they are located within 50 miles of home.
  - By some calculations, the number of genetic providers in the state is little more than one-third the amount needed to provide appropriate care for Virginia's current population.
  - Many of Virginia's PCPs identified access to genetic services as a major problem for a significant number of their patients.
  - The majority of stakeholders whom have say in the development of policy and regulations regarding genetics, in the determination of coverage for services, and the potential treatment of patients are uninformed about genetic care.
  - Growing linguistic and cultural diversity in the patient population present challenges to existing genetic resources.

- Funding gaps present a major impediment to genetic services in Virginia.
  - Clinical genetic care is usually very time-consuming, with a large, labor-intensive cognitive component and an emphasis on preventive healthcare, which has historically not been well reimbursed by insurers.
  - Most insurers now cover genetic consultations but may not cover genetic testing. Most reimburse at fairly low levels that may not allow out-of-state genetic testing to be performed, even when in-state testing may be unavailable.
  - Surveyed genetic providers cited poor funding, limited insurance coverage, and inadequate reimbursement as the most significant barriers facing genetic services in Virginia.
  - Surveyed PCPs indicated that the cost of genetic services prevents a significant proportion of their patients from obtaining care.
  - Genetic providers reported that insurance reimbursement pays for approximately half of the actual cost of providing genetic care on average.

- Limited knowledge and awareness of genetics and genetic services among non-genetic healthcare providers, key stakeholders, and the public are a growing concern.
  - In the future, PCPs and non-genetic specialists will increasingly be called upon to provide some genetic services and will be required to function as informed gatekeepers of genetic healthcare.
  - Multiple studies nationwide have documented chronically inaccurate genetic risk assessment, inappropriate testing, and misinterpretation of genetic test results by PCPs, non-genetic specialists, and allied health professionals.
  - Despite significant media attention and multiple educational initiatives, the few studies available indicate the general public has a limited understanding of genetics, along with some misconceptions about heredity.
  - The majority of genetic providers surveyed indicated that Virginia's non-genetic healthcare professionals and the public are not sufficiently knowledgeable about genetics or available genetic services.
  - The majority of the general public and key stakeholders surveyed rate their own level of genetic

knowledge as average or below.
  - Key stakeholders rated the average Virginian's genetic literacy as poor to very poor.
  - Virginia's PCPs, consumer focus group participants, key stakeholders, and the general public identified education about genetics and genetic services as one of the most crucial needs currently facing genetic healthcare in Virginia.
  - Despite this need, most of Virginia's genetic providers, particularly those in non-academic settings, have limited involvement with educational initiatives targeting the public or non-genetic healthcare providers.

- There is an ongoing need for attention to the ethical, legal, and social implications of genetics.
  - Some state and federal laws regarding genetic discrimination and privacy are in place.
  - The majority of surveyed genetic providers are not satisfied with the protection afforded by existing legislation in Virginia.
  - Studies of non-genetic healthcare providers indicate that the practice patterns of these providers are often not conducive to genetic privacy, nondiscrimination, or informed consent.
  - Key stakeholders identified a need for more public discussion of ethical, social, and legal issues as a top future need for genetic services in Virginia.

- Improved linkages between different genetic services and databases in the state as well as with other medical specialties, such as epidemiology and adult chronic disease services, are needed.
  - A growing understanding of genetics and its applications throughout medicine is resulting in expanding needs for genetic services in oncology and other adult specialties.
  - The Centers for Disease Control and Prevention (CDC), CORN, and other national public health genetics groups have stressed the benefits of linkages between genetic services and other medical specialties, as well as links between genetic data sources.
  - Virginia's genetic consumer focus group participants identified coordination of care between genetic providers and other specialists caring for individuals with genetic conditions as a critical need.
  - Improved coordination of services was noted as a future need by key stakeholders.
  - National studies have documented a significant lack of awareness of genetic services among adult chronic disease specialists, a trend echoed by a small pilot project in Virginia.
  - Virginia lacks cancer genetic services in multiple regions, while other adult genetic services are largely non-existent.
  - Although several efforts to enhance linkages between genetic data sources are underway in Virginia, further improvements would be beneficial.

While Virginia has a solid foundation of genetic services, gaps remain. Other states and national organizations have developed programs and initiatives designed to meet needs in genetic services and education that can serve as models for Virginia. Optimally, future planning will include the development of a comprehensive state genetic plan that is designed to meet the needs of Virginia's residents and incorporates the insights obtained through these data collection and community assessment processes.

# 1. AN INTRODUCTION TO GENESEAN

## Medicine and the New Genetics

After decades of research through programs such as the HGP, most diseases, from rare birth defects to common adult conditions, are now known to have a genetic component. Collectively genetic conditions are the principle cause of infant death, a major contributor to childhood hospitalization, the main cause of pregnancy loss, and a central determinant of premature adult death. It is estimated that 18-30% of children have a chronic physical, developmental, behavioral, or emotional condition, most of which are of inherited etiology. Children with special health care needs, many with genetic conditions, account for approximately 80% of child health expenditures yearly. Unfortunately, the United States (U.S.) has the worst infant mortality rate among the G-7 industrialized nations and ranks 25$^{th}$ internationally, principally as a result of birth defects and related conditions (1). These statistics illustrate the relevance of genetics to medicine and highlight the possibilities for improving health outcomes for all Americans.

As scientists continue to unravel the mysteries of genetics, further advances in the diagnosis, treatment, and, eventually prevention of many disorders will become possible, presenting both incredible opportunities as well as multiple challenges to the current health care system (2). Genetics is becoming an increasingly fundamental component of medical practice and healthcare policy, challenging healthcare providers and policymakers in Virginia and elsewhere to integrate genetics across health systems. It is widely recognized that accessible, affordable, culturally appropriate genetic services as well as improved genetic literacy of the American public need to be in place for the fruits of the HGP to be realized (1). The realities of birth defects prevention and improved genetic health require an adequate number of providers, a system of appropriate triage and referral, and ongoing attention to identified gaps in education and service. To address these growing opportunities and challenges, the federal Maternal and Child Health Bureau (MCHB), the CDC, CORN, and many other organizations have indicated the importance of a statewide genetic plan, while also stressing that a needs assessment is a critical first step in developing a comprehensive plan. Several states have already conducted needs assessments and developed state genetic plans, and CORN has provided a framework for conducting needs assessments (3). Recognizing the importance of conducting long-range planning to translate genetic discoveries into improved health outcomes, the Virginia Genetic Advisory Committee (VaGAC), composed of state leaders from genetic-related agencies and organizations and convened under the leadership of VDH, has initiated the development of a state genetic plan for Virginia. To facilitate informed planning by state leaders and policymakers, the VCU Department of Human Genetics, with funding from the Virginia Chapter of the MOD, has undertaken a comprehensive genetics needs assessment project, *Virginia GeneSEAN: Genetic Services and Education – An Assessment of Needs*, the results of which are presented in this report.

## GeneSEAN Project Overview

The ultimate goal of the Virginia GeneSEAN project is to assure that high quality systems of care and interventions are available so that all Virginians can access appropriate care for genetic health concerns. Under this ideal, the purpose of the project is to document the current status of medical genetic services and education throughout Virginia, specifically addressing existing systems of care, prevention services, educational activities, insurance issues, data collection, and QA, as well as gaps in these systems. This report is intended to provide a framework for targeted planning and service initiatives, educational interventions, and systems development to address gaps and disparities identified by the needs assessment. The target audience for this needs assessment report includes health planners, state agencies, insurers, legislators, health professionals, funding organizations, and other policymakers and

stakeholders, who are encouraged to use the information and data contained in the report in planning for effective resource allocation, improved health, reduced health disparities, and other efforts aimed at improving the state of genetic healthcare and education in Virginia. Needs assessments in other states have had impacts on QA, funding of clinical services infrastructure, educational activities, database integration, and increased access through enhanced provider and insurer awareness. It is hoped that this assessment will be used similarly in Virginia.

To accomplish the goals of Virginia GeneSEAN, the project team focused on the following main objectives:

- Describe the current status of genetic services and education in the state as well as barriers to high quality and appropriate community-based, family-centered genetic care and information by reviewing existing data sources.
- Assess the current and emerging practices and gaps within genetic health services and education delivery in Virginia through consumer focus groups and surveys of genetics professionals, PCPs, insurers, VDH staff, and other stakeholders.
- Obtain the participants' input on ways to improve the existing system and address the current and growing disparities in access to genetic services, education, and testing.

Several activities have been undertaken over the past three years to meet these objectives. In 2003, a comprehensive review of the medical literature and other relevant data sources was performed, focusing on topics related to the history of genetics, Virginia health statistics, existing genetic service systems in the state, non-genetic healthcare provider roles, the status of professional and public genetic education, and fiscal issues affecting genetic services. Simultaneously, a detailed survey was developed with the assistance of the VaGAC and VDH and distributed to all identified clinical genetic providers throughout Virginia. The survey addressed practice patterns and service provision, as well as providers' perceptions of a variety of factors, including the quality and accessibility of genetic services in Virginia, availability of resources, reimbursement issues, legal and ethical issues, the role of state organizations, and educational needs and practices. In 2004, a brief survey assessing PCPs' perceptions of genetic services and education was developed and distributed to over 5000 PCPs and health department district directors throughout the Commonwealth. An assessment of genetic consumers' views regarding the quality and accessibility of genetic services in Virginia, genetic services needs, educational needs and practices, and genetic discrimination and privacy was also undertaken in 2004 through the development of a focus group tool that was used to conduct consumer focus groups in two regions of the state. The findings of these activities have been analyzed and are summarized in this report. The CORN assessment guidelines are used to illustrate gaps and highlight successes in Virginia's systems of care, and insights gained from an analysis of other states' genetic needs assessment reports, genetic plans, and resulting initiatives are included as well. In 2005, in order to provider a more comprehensive view of genetic needs, further input was solicited through a survey of other identified key stakeholders. These stakeholders included representatives of insurance companies, specialty doctors (i.e. oncologists, surgeons, cardiologists), allied healthcare professionals (i.e. nurses, nurse practitioners), and major employers in Virginia; representatives of regional hospitals, medical schools, and organizations; government representatives; and representatives from the education system and the media. Finally, a Commonwealth Poll was conducted in 2005 to gather the opinions and viewpoints regarding genetic services of the citizens of Virginia.

This report is the final publication that incorporates the results of all of the activities conducted over the three-year period. It is expected that this final document will not only serve as a resource for the development of a state genetic plan, but that it will also be useful for private and public agencies concerned about quality services and access in maternal and child health, advocacy groups concerned about genetic services and literacy, and individuals and families who live the stories behind the statistics of health disparities in Virginia.

## 2. HISTORICAL BACKGROUND

"Youth, thou bear'st thy father's face;
Frank nature, rather curious than in haste,
Hath well compos'd thee. Thy father's moral parts
Mayst thou inherit too!"
~ *All's Well That Ends Well* by Shakespeare

Nearly every day, newspaper headlines report the discovery of a new gene implicated in human disease. Yet in 1953, when James Watson and Francis Crick reported the elucidation of the structure of deoxyribonucleic acid (DNA), their discovery was hardly considered newsworthy. In the 50 years since this discovery, basic genetic research has made remarkable progress, with innumerable applications to medicine. Gregor Mendel, considered the father of genetics, could not have dreamed of such a heritage, but his pea plant experiments were a hallmark event along the circuitous route that led to the sequencing of the human genome. Mendel's theories of inheritance provided the framework on which today's genetic medicine is based. Along the way, Hermann Muller demonstrated that genetic mutation was more than just an idea, Linus Pauling found the link between gene mutation and human disease, and Avery, McCarty, and MacLeod proved that DNA was the material of inheritance. Rosalind Franklin's X-ray crystallography images facilitated Watson and Crick's construction of the double helix model, and innumerable others provided clues that have facilitated the unraveling of the mysteries of genetics (1). Today, such discoveries as imprinting, transposons, and trimethylation repeats continue to unfold.

The examination of family history information is a cornerstone of 21$^{st}$ century medical practice, particularly in clinical genetics. In the same way, a complete understanding of the current practice of clinical genetics requires an examination of *its* history, including not just a chronology of events, but also a tracing of the thoughts that evolved with time into the current perception of the field. The themes of today are conditioned by the past, thus an informed plan for the future requires an understanding of the flow of past ideas that have led to the current state.

### Ancient History: Laying the Foundation

The individuals considered the founding fathers of genetics were by no means the first to ponder the secrets of heredity. As evidenced by Shakespeare's quote above, the idea of inheritance, albeit by an unknown mechanism, was a part of everyday life for many centuries before Mendel (2). Earlier historical evidence confirms a long-standing interest in variations of the human form. Statues of individuals with achondroplasia have been found in the Egyptian pyramids, and descriptions of dwarfing syndromes can be found in 4000-year-old writings from the First Babylonian dynasty (3). Hemophilia, although named otherwise, was mentioned in the Talmud and was known in the Middle Ages as "passio flux sanguinis" (4). Archeologists have found evidence of various congenital anomalies in ancient Greek, Roman, Japanese, and other cultures. Greek writings of the fourth century B. C. and earlier describe genetic concepts, presumably reflecting popular perceptions of that era as well as the theories of the physicians and philosophers of the time. While these writings are not based on scientific evidence, they illustrate the ideas that undoubtedly served as the foundation on which the ancient philosophers and physicians of the Hippocratic school developed their theories of heredity, which influenced the development of genetics and medicine, indeed all of science, for many centuries (5).

## The Birth of Genetic Science

Hippocrates suggested that the material of inheritance consisted of particles from throughout the body that had been shaped by experience, while Aristotle believed that the instructions were constant and contained in the gametes. Such speculations remained the province of philosophers until the microscope was invented by Anton van Leeuwenhoek in the 17th century. Economic support for genetic research came from an unexpected quarter at this point, as international trade expanded in the Age of Discovery and the Old World was introduced to many new plant species. Improvement of plant cultivars and animal breeding experimentation became a driving force behind much of the scientific research in genetics.

The city of Brunn in Moravia, now Brno, Czech Republic, was a center of the textile industry of the early 19th century. Its leaders were interested in increasing economic profits by improving sheep breeds and thereby enhancing wool production, and organized societies to promote scientific research in an effort to facilitate this goal. In this atmosphere of scientific agricultural research, Gregor Mendel, who had studied physics in Vienna previously, took a quantitative approach to his pea breeding experiments, drawing on his physics training. His techniques allowed for the formulation of a mechanical description of the laws of heredity. He proposed that hereditary information was passed from parent to offspring through units he termed "factors." Of critical importance was his suggestion that these factors were inherited in pairs, one from each parent. He also recognized that one factor might dominate over the instructions of its partner in determining the resulting trait, with the other factor remaining in latent form to be passed to subsequent generations, where its effects could reappear in predictable ratios. Mendel reported his findings in 1865 in the *Journal of the Brunn Society of Natural Science*. Unfortunately, his results were based on mathematical formulas and unseen "factors," and remained largely unknown to the world of biology during his lifetime.

By the middle 1880s, improved microscopy led researchers to recognize that the physical unit of inheritance was contained in the nucleus and that the egg and sperm seemed to contribute equally to heredity. Chromosomes, easily visible components of the nucleus, were an obvious candidate as the facilitators of inheritance, however, many researchers continued to believe that the cytoplasm was the unit of inheritance. In early 1900, three researchers -- Hugo de Vries, Erich von Seysenegg, and Carl Correns -- independently "rediscovered" Mendel's laws and reported them to a scientific audience that by then had enough background knowledge to recognize the fundamental importance of those findings.

Research in the early 20th century then focused on finding Mendel's "factors" in the cell nucleus. The study of the fruit fly *Drosophila* emerged as a critical element in the unraveling of the genetic mystery, with researchers in Thomas Hunt Morgan's lab at Columbia University, including Alfred Sturtevant, leading the way. He realized that a linear "linkage map" of genes, as Mendel's factors came to be called, explained the co-transmission of traits and hypothesized that those genes were lined up along the threadlike chromosomes.

In 1927, Hermann Muller demonstrated that radiation could mutate *Drosophila* genes, providing further proof that genes were physical units within the cell. At the Rockefeller Institute in New York, Oswald Avery, Colin McLeod, and Maclyn McCarty worked on isolating the elusive genes, eventually providing evidence that DNA molecules from bacteria were capable of transmitting genetic instructions. Some researchers continued to believe that DNA was merely a structural component of chromosomes and that proteins were the actual unit of heredity. By the early 1950s, however, further experiments had proven the skeptics wrong, yet the baffling question of *how* DNA encoded hereditary instructions still remained.

At Cambridge University, James Watson and former physicist Francis Crick, relying on critical X-ray

crystallography work by Rosalind Franklin, finally elucidated the molecular structure of DNA in April 1953. Their double helix DNA model composed of four nucleotide bases explained how genetic information was encoded, how it could be copied, and how mutations could arise. By 1964, researchers had learned how the genetic code could be translated into a messenger ribonucleic acid (RNA) molecule that subsequently directed the synthesis of a specific protein from amino acids. The ability to "read" the sequence of an actual gene did not come until the 1970s, however, when the Sanger and Maxam-Gilbert techniques for DNA sequencing were independently developed.

The ability to sequence genes led to an explosion of knowledge about the nature of heredity. Continual improvements in sequencing technology hastened the pace of discovery. The systematic study of complete genomes, a concept termed "genomics," was initiated by a process involving an extension of the idea of genetic mapping originally proposed by Sturtevant in the 1920s. David Botstein suggested that a complete genetic map of the human chromosomes could be constructed by association with the inheritance of DNA polymorphisms, or common DNA sequence variations. Each polymorphism could be localized to a specific site on a specific chromosome. By matching the inheritance of specific polymorphisms with a genetic disease within a family, one could localize the gene causing the disease. This strategy proved successful in the hunt for the gene causing Huntington disease, which was mapped to the short arm of chromosome 4 in 1983. The revolution of clinical genetics had begun, as hundreds of disease genes were mapped shortly thereafter. By 1985, the HGP, with its seemingly impossible goal of sequencing the entire human genome, had been envisioned (6).

## A Closer Look: The Influence of the Eugenic Movement

The term "eugenics" was popularized by Sir Francis Galton, considered the founding father of quantitative genetics and eugenics, as well as being the biological cousin of Charles Darwin. Galton, a contemporary of Mendel, studied multifactorial inheritance and endeavored to find statistical methods to explain the complex inheritance of such traits as intelligence, stature, eye color, and liability to infectious disease. To accomplish this, he collected extensive family medical records, including twin studies, and developed robust statistical measures for data analysis (7). Simultaneously, he became involved with and eventually championed the theories of eugenics, which he defined as the "science of improving heredity." (8) Advocates of eugenics thought that the future of the species depended on eradicating disease, including mental, social, and physical illness, much of which was thought to be hereditary. Efforts to accomplish this included immigration restriction, sterilization and antimiscegenation laws, and eventually, in Nazi Germany, genocide (7).

Unfortunately, many of the first human geneticists and human genetics research institutions of the early 20$^{th}$ century became entangled in the emerging eugenic sociological theories. Support for human genetics research at that time continued to come from organizations with roots in plant and animal breeding, which likely facilitated the focus on species improvement as a goal in humans as it had been in plants and animals. In fact, the American Breeders' Association changed its name to the American Genetics Association in 1914, and its main publication, the *American Breeders Magazine*, became known as *The Journal of Heredity.* Alexander Graham Bell contributed the lead article of the new journal, entitled "How to improve the race." Studies produced by the Eugenic Record Office, founded in New York in 1910, included some that attempted to demonstrate that insanity and criminality were simple Mendelian traits. A 1914 report lists 44 universities with courses in genetics and eugenics, offered as often by sociology departments as by biology or zoology departments (9). Eugenics also provided a medicalization of pre-existing racism, as exemplified by the Virginia Racial Integrity Act, a law passed in 1924 that prohibited the marriage of Caucasians to non-Caucasians. Such laws were in place in many states, and variably enforced, until the Virginia law was struck down by the U. S. Supreme Court in 1967 (10).

Virginia's role in the eugenic movement was also cemented by the 1927 Supreme Court case *Buck v Bell*, in which the court ordered that surgical sterilization of Carrie Buck, a young woman diagnosed with "hereditary feeblemindedness" and a resident of a Virginia mental institution, was constitutional (11). An estimated 60,000 legal sterilizations reportedly took place in the United States in the first half of the 20th century under similar sterilization laws (12).

Concerns about the discrimination inherent in the eugenics movement have impacted the current practice of clinical genetics significantly, from non-directiveness as a tenet of genetic counseling to the focus on ethical and social issues. Leaders in the field explicitly renounce eugenics, and significant investments are put toward studying these issues, as evidenced by the HGP's *Ethical, Legal and Social Issues Branch* (13).

## Of Fruit Flies and Man: The Origins of Clinical Genetics

Prior to a recognized field of clinical genetics, numerous physicians and medical researchers contributed to the body of knowledge on which the field developed by publishing their findings in various medical or natural history journals, as no journals of heredity or genetics existed before the 20th century. Maupertuis described an autosomal dominant postaxial polydactyly in four generations of a family in 1752; Bemiss reported on the effect of consanguineous marriages on resulting offspring in 1858; Lyon described what is now known as Huntington disease in 1863; and Alexander Graham Bell pointed out the role of heredity in deafness before 1888 (14, 15, 16). For generations, physicians and medical researchers have been intrigued by hereditary disease, yet the practice of clinical genetics was not an organized entity or really a coherent idea until well into the 20th century. Victor McKusick, founder of the Division of Medical Genetics at Johns Hopkins University, marks the origins of clinical genetics to the 1950s, when findings in cytogenetics and biochemical genetics combined with the progress of basic genetics to form a critical mass of information that began to have a significant impact on the practice of medicine. Research about blood groups, clotting, and disorders of the eye and skin contributed to this critical mass as well (17).

Walther Flemming, an anatomy professor in Kiel, Germany, first described chromosomes in 1876. However, advances in cytogenetics were initially limited, with primitive technology leading to an incorrect assessment of the number of human chromosomes by T. S. Painter (18). Finally, the technique of tissue culture of human cells allowed Tijo and Levan to correctly identify the number of chromosomes in 1956 (19). In 1959, the chromosomal etiologies of Down syndrome, Turner syndrome, and Klinefelter syndrome were revealed, although the disorders themselves had been described much earlier. By this time, refinements in techniques for studying chromosomes made the practice of chromosome analysis from a blood sample a fairly simple procedure, facilitating the rapid identification of many other human disorders of chromosomal etiology (17).

Archibald Garrod, a clinical rheumatologist, is considered the father of biochemical genetics. In the course of his clinical practice, he encountered and subsequently described the disorder of alkaptonuria, as well as albinism and cystinuria, as Mendelian autosomal recessive traits in 1902 (20). The field of biochemistry began to blossom in the early 20th century along with an understanding of inborn errors of metabolism as researchers refined techniques and demonstrated the workings of various enzymes in vitro. By the 1950s, biochemists had developed chromatography techniques for screening urine for abnormal metabolites, the possibility of therapy for phenylketonuria (PKU) was realized, and techniques for hemoglobin electrophoresis and starch gel electrophoresis were developed. All of these advances simplified the process of searching for structural abnormalities in enzymes and other proteins (17). By the early 1960s, population-wide newborn screening programs for PKU were initiated and later extended to other inborn errors of metabolism. These population-based programs contributed to the growth of

mathematical and population genetics, and to their integration into the practice of clinical genetics. Immunogenetics expanded to include a whole realm of disorders of the immune system, which began to come to light only after the antibiotic era. Agammaglobulinemia, for example, was not recognized until 1952, because earlier cases of this and other immune deficiency disorders were unable to be differentiated from the many casualties of childhood infectious disease before the widespread availability of antibiotics (17).

Interestingly, the origins of clinical genetics are solidly founded in basic science, while other specialties of medicine originated largely as crafts, with only a retrospective study of the basic science. An examination of papers published in early genetics journals reveals that much more effort was targeted toward the study of basic genetics than human genetics during the first decades of the 20th century (9). The exceptions often came from those same basic genetics researchers; however, perhaps contributing to the strong link between research genetics and clinical genetics, a trend that remains today. In 1925, H.G. Muller, a leading Drosophila geneticist, published work on the inheritance of mental traits obtained from a study of identical twins (21). Following this publication, twin studies became more popular as a research methodology and contributed to the rise of human genetics as a viable research area. Because clinical genetics did not exist as a field of study at the time, the first practitioners of clinical genetics came from other fields. A handful were physicians and dentists interested in human genetics, such as William Allan of North Carolina, who published a classic paper on the inheritance of migraine in 1928 and later developed the human genetics program at Bowman Gray School of Medicine. Some were Drosophila and other animal geneticists who gradually became interested in human traits and moved their research in that direction. Gardner syndrome, for example, was the first human disorder named after a person with a Ph.D. and not an M.D. degree. By the 1950s, many more physicians trained in a wide range of fields began to work on human genetic diseases.

By the late 1940s, there were centers of research in human genetics at several universities in the U.S., as well as a critical mass of individuals interested in human genetics. At a meeting of the American Association for the Advancement of Science in 1948, the American Society of Human Genetics (ASHG) was established with Drosophila geneticist H. J. Muller elected president. The first issue of the American Journal of Human Genetics was published in the fall of 1949. (9) The creation of a new specialty of clinical genetics was underway, as clinical genetic clinics were created or evolved from human genetics research groups, with the charge of caring for patients with "genetic" diseases. These were usually divisions within pediatric, internal medicine, or obstetric departments, although a few were independent genetic departments (22). The first of these clinical units was established at Ohio State University in 1934, followed by programs at Bowman-Gray, the University of Michigan, the University of Minnesota, and the University of Utah in the 1940s (23). By 1975, there were 91 departments or divisions of medical genetics in the U.S. (22).

For clinical genetics, the 1960s and 1970s were largely an era of syndrome delineation, with the role of the clinical geneticist as dysmorphologist, an expert in rare disorders and observational skills, gradually established in the medical community (24). For many years, genetic counseling was largely provided by the basic science geneticists that had crossed over into human genetics, as there were few physicians with the requisite knowledge or interest. The medical community gradually took over this role; although some overlap remained into the 1970s at least. Around the same time, a new group of genetic professionals emerged, originally termed genetic associates and later genetic counselors, as providers of genetic counseling and integral members of the clinical genetics team (25). The first formal training program for genetic counselors was initiated at Sarah Lawrence College in New York in 1969, with the first master's degrees issued to eight women in 1971. The term "genetic counseling" was initially proposed by Sheldon Reed as a replacement for "genetic hygiene," with its unsavory eugenic ties. The modern definition

conceptualizing genetic counseling was developed based on discussions held at a 1972 "Workshop on Genetic Counseling" sponsored by the National Genetics Foundation:

> Genetic counseling is a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family 1) comprehend the medical facts, including the diagnosis, the probable course of the disorder, and the available management; 2) appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives; 3) understand the options for dealing with the risk of recurrence; 4) choose the course of action which seems appropriate to them in view of their risk and their family goals and act in accordance with that decision; and 5) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder (26).

Clinical genetics gradually became recognized as a medical subspecialty, with the 1980 founding of the American Board of Medical Genetics (ABMG) as the certifying board for Ph.D. and M.D. geneticists and genetic counselors. The American Board of Genetic Counseling (ABGC) was later formed and took over the task of certifying genetic counselors as of 1993, which facilitated the recognition of ABMG by the American Board of Medical Specialties (ABMS) in 1991 (27, 28, 29).

A genetics presence was established in Virginia in 1938, when the School of Pharmacy at the Medical College of Virginia (MCV) hired Drosophila geneticist Roscoe Hughes to teach biology and anatomy. Dr. Hughes was gradually able to introduce genetic courses into other MCV curricula, including what was probably the first dental genetics course ever, initiated in 1949 and formalized in 1954, as well as medical genetics in the School of Medicine in 1951. The first graduate degree in genetics was awarded by the department in 1953, while the first genetics clinic was held in the late 1960s. In 1975, the university implemented a plan to expand the program with the recruitment of Walter Nance as chairman of the renamed Department of Human Genetics, which was soon followed by the hiring of additional faculty as well as a significant increase in research activities (30, 31). A master's degree program in genetic counseling was initiated at MCV in 1990. The program has since graduated more than 50 genetic counselors, many of whom work throughout the state (32). Meanwhile, Meinhard Robinow and Thaddeus Kelly established a division of medical genetics at the University of Virginia (UVA) in Charlottesville in 1975 and quickly set up a system of satellite clinics throughout western Virginia and southeast West Virginia (33). The Children's Hospital of The King's Daughters (CHKD) in Norfolk established a division of medical genetics in 1988, which has since provided genetic services throughout eastern Virginia and northeastern North Carolina in conjunction with other specialized genetics services provided by its partners at the Eastern Virginia Medical Center, including infertility and preimplantation genetics at the Jones Center and obstetric genetics through Eastern Virginia Medical School (EVMS) (34, 35). In northern Virginia, Genetics and IVF (GIVF), founded in 1984 by Dr. Joseph Schulman, is a private facility providing genetic services as well as all aspects of infertility treatment, including in vitro fertilization and preimplantation genetic diagnosis (36). A handful of other clinics, staffed by genetic counselors providing genetic services independent of the main centers, have gradually been established throughout the state over the last 30 years.

### The View from Outside: The Integration of Genetics into Other Medical Specialties

Although every field of medicine involves hereditary disorders and genetic issues, clinical genetics specialists have historically worked most closely with pediatricians and obstetricians. Pediatricians are

usually the main healthcare providers for infants and children with as-yet-undiagnosed genetic syndromes, which are often quite subtle initially and are frequently rare. Pediatricians have generally come to rely on geneticists as experts in dysmorphology and syndrome identification for assistance in the diagnosis and management of these patients (3). In obstetrics, the links to clinical genetics were largely established by the cytogenetic discoveries of the mid-20th century combined with the development of techniques for amniocentesis in the 1950s. Those discoveries, along with several lawsuits in the 1970s in which parents of children with disabilities sued their obstetricians for failing to refer them for amniocentesis, led to the widespread use of prenatal genetic testing (37). Genetic disease overall has continued to rise as a proportion of the total disease burden in industrialized nations because many other forms of disease, such as infectious and nutritional disease, have come under better control in the 20th century. This fact, combined with increasing discoveries about the role of genetics in all disease and the proliferation of genetic testing technologies, has resulted in a shift away from the perception that genetic disease is limited to rare conditions seldom encountered by clinicians (13). Increasingly, all medical specialties are finding that a broad knowledge of genetics is a requirement of good clinical practice (38).

## The Next Frontier: The Human Genome Project

The idea of reading the entirety of the human genome's 3 billion bases of DNA seemed like a foolish waste of time and money to some, particularly because much of it was thought of as "junk" DNA, spacer DNA sections that had no significant function. Discussions proceeded, however, and the HGP was officially launched in 1990. The effort was an international one, with hundreds of scientists at 20 sequencing centers in China, France, Germany, Great Britain, Japan, and the United States contributing to the project. In the late 1990s, the effort was further accelerated when Celera Genomics, a privately funded organization, announced a competitive effort to sequence the genome using a different strategy (39). The successful completion of the HGP was officially announced in April 2003, the 50th anniversary of *Nature*'s publication of Watson and Crick's landmark paper describing the structure of DNA. The first draft of the sequence was announced in June 2000, while the more precise finished sequence covers about 99% of the human genome's gene-containing regions and has an accuracy of 99.99%. The original plan called for a completion date of 2005 with a budget of $3 billion in fiscal-year 1997 dollars; however, not only was the sequence completed well ahead of schedule, it also cost less than originally expected (40).

Although conflicting views about the patenting of DNA sequences are still a significant issue in genetic research today, a hallmark feature of the HGP was the 1996 adoption of the so-called "Bermuda Principles," which required automatic, rapid release of sequence data to the public domain, allowing scientists worldwide to immediately use sequence information. This has resulted in an ever-increasing pace of discovery in genetics, including an increase in the number of identified disease genes from less than 100 prior to the HGP to over 1400 by its completion. The newfound understanding of the genome is expected to accelerate research into the causes of disease as well as approaches toward prevention and treatment. With the completion of the HGP, the National Human Genome Research Institute (NHGRI) is now focusing on research promoting application of the new knowledge, including:

- The development of technologies to facilitate identification of the hereditary contribution to common diseases such as diabetes, heart disease, and mental illness;
- Better methods of early disease detection;
- Economical technologies for whole genome sequencing, aimed toward eventual incorporation into everyday medical practice; and
- Wider application of "chemical genomics" to increase the understanding of biological pathways and accelerate drug discovery (40).

## A Glimpse into the Future

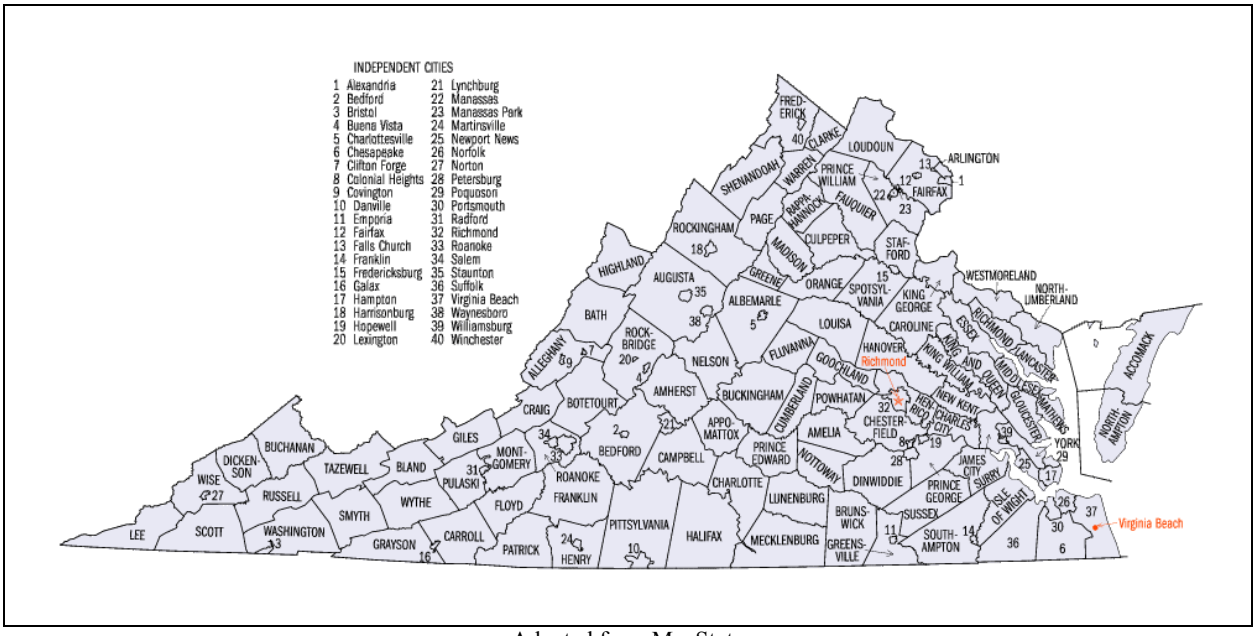
With the plethora of information provided by the realization of the goals of the HGP, 21st century biology is shifting its focus yet again. Future goals include creating a catalog of all the common variants in human genes and finding associations with disease states, a particularly intriguing possibility because common variants are thought to be involved with susceptibility to common diseases. The post-genome era will involve functional genomics, or the elucidation of the functions of genes, as well as proteomics, the determination of the function of proteins (39). After decades of reductionist methods, scientists are moving toward more holistic views of biological systems. As succinctly stated by Francis Collins, NHGRI director, the genetic revolution in medicine is under way (41).

## 3. WELCOME TO VIRGINIA: A LOOK AT THE STATE AND ITS HEALTH

The Commonwealth of Virginia, one of the original 13 colonies of the United States, was the site of the first permanent English settlement in America and England's first successful overseas colony. Nicknamed the Old Dominion, Virginia played an important part in the American Revolution, helped shape the democratic principles on which the country was founded, and served a critical role in the American Civil War, when the state's capital, Richmond, was also the Capital of the Confederacy. While undoubtedly shaped by her storied history, Virginia today must plan for the future prosperity of her citizens, including their health. Overall, Virginians tend to fare well on national measures of health status, particularly compared to other southern states, with average to above average rankings on most criteria.

### Virginia's Geography

**Figure 3-1. Virginia - Counties and Independent Cities**




Adapted from MapStats

Virginia covers 42,326 square miles (109,624 km$^2$), including 2728 square miles (7065 km$^2$) of inland and coastal waters. The state is roughly triangular in shape and is bounded on the east by the Atlantic Ocean, on the north and east by Maryland and the District of Columbia, on the west by West Virginia and Kentucky, and on the south by Tennessee and North Carolina. A map of the state indicating all counties and independent cities is shown below. Five natural geographic regions extend across the state, including the Coastal Plain in the east and four subdivisions of the Appalachian region and the Appalachian Mountains. Multiple rivers cross the state, including many that empty into the Chesapeake Bay, which divides the southern tip of the Delmarva Peninsula, or Eastern Shore, from the state mainland, and is one of the world's largest estuaries. Virginia's coastline is 112 miles (180 km) long, while the tidal shoreline measures 3315 miles (5335 km). Forests, mostly deciduous hardwood, cover 63% of the state. The climate is temperate, with hot, humid summers and mild, wet winters, although temperatures vary significantly across the state. Air pollution is a concern in Northern Virginia, part of the large urban population surrounding Washington, D.C. The Chesapeake Bay has been damaged by runoff from agriculture and coal mines as well as urban and industrial pollution, and is the focus of a cooperative

clean-up effort among multiple states bordering the bay. Virginia had 30 hazardous waste sites on a national priority list in 2003 due to their severity or proximity to population centers. Progress in pollution reduction efforts has been made, however, with a 10% reduction in toxic chemicals released to the environment over the period from 1995 to 2000 (1). These environmental issues can have a significant impact on the health of Virginians, including the incidence of birth defects and the morbidity of genetic conditions.

## Economic Foundations

Prior to the Civil War, the economy of Virginia largely depended on tobacco farming; however, after the war, agriculture in the state was increasingly centered on diverse products, including livestock and grain. Various industries developed during the 20th century, including coal mining in the southwestern part of the state. During and after World War II, there was a huge expansion of the shipbuilding industry in southeastern Virginia, an area known as Hampton Roads, as well as significant growth in federal government activity in the Washington D.C. area, including the region now known as Northern Virginia. Tourism has gradually become more important to the state economy as well. These economic developments resulted in an increasingly urban population in the state, and increased the standard of living so that by 1990, Virginians' per capita income was greater than the national average and was the highest among southern states. Income within the state varies tremendously, however, with very low levels in the far southwest and very high levels in Northern Virginia (1). The 1999 poverty rate for all ages was 9.6% in Virginia, with 12% of Virginia's children living in poverty (2, 3). Obtaining healthcare remains a hurdle for these families. In December 2002, more than 300,000 children in Virginia were enrolled in Medicaid or Family Access to Medical Insurance Security (FAMIS), the state-sponsored health insurance programs, however, an estimated 98,000 eligible children were still uninsured (4, 5).

In 2002, Virginia's labor force included 3,735,000 people: 40% worked in services, 23% in wholesale or retail trade, 12% in manufacturing, 6% in government (including the military), 6% in construction, 6% in transportation or public utilities, 5% in finance, insurance, or real estate, 1% in farming, forestry, or fishing, and 0.3% in mining. Although farming accounts for a small proportion of the workforce, it remains a significant part of the state's economy, with over 1/3 of the state's land area covered in farmland. Livestock and livestock products, particularly poultry, are the leading source of agricultural income, while tobacco farming is second. Virginia is also among the top ten states in commercial fisheries, particularly shellfish. The primary manufacturing sectors in Virginia are the production of chemicals and associated products, the processing of agricultural goods from Virginia's farms, and the production of transportation equipment, including shipbuilding and ship repair. Transportation contributes to the economy significantly, with Hampton Roads being one of the country's leading seaports (1).

Although economic growth in Virginia has been significant since World War II, it has mostly benefited Northern Virginia and Hampton Roads. Other areas of the state, particularly the rural Southwest, the Eastern Shore, and areas south of Richmond, have lagged behind. Suburbs are prospering while inner cities are plagued by unemployment, crime, drugs, and broken family structures, echoing similar trends throughout the country. Long-standing industries in the state, particularly mining, commercial fisheries, and tobacco farming and cigarette production, face uncertain futures.

## Who is Virginia?

The population of Virginia was slightly over 7 million according to the 2000 census, an increase of 14.4% compared to 1990. Census projections estimate a population of approximately 8 million by 2015 and 8.5 million by 2025 (6). Residents of urban areas comprised 73% of the population, a number that has been

steadily rising since World War II. A metropolitan corridor, containing over half of the state's population, has developed in the area from Northern Virginia south to Richmond and east to Hampton Roads. Table 3-1 shows the population in the state's metropolitan areas, while the population in the state's ten largest cities is shown in Table 3-2. Several urbanized counties also have large populations, notably Fairfax (969,749), Prince William (280,813), Arlington (189,453) and Loudoun (169,599) in Northern Virginia and Henrico (262,300) and Chesterfield (259,903) in the Richmond metropolitan area (7).

**Table 3-1. Virginia's Metropolitan Area Populations (2000 census)**

| Metropolitan Area | Population |
|---|---|
| Bristol* | 91,873 |
| Charlottesville | 159,576 |
| Danville | 110,156 |
| Lynchburg | 214,911 |
| Norfolk-Virginia Beach-Newport News* | 1,551,351 |
| Northern Virginia* | 2,167,757 |
| Richmond-Petersburg | 996,512 |
| Roanoke | 235,932 |

*Population figures include only the Virginia portion for metropolitan areas that extend into neighboring states.

**Table 3-2. Virginia's Ten Most Populous Cities (2000 census)**

| City | Population |
|---|---|
| Virginia Beach | 425,257 |
| Norfolk | 234,403 |
| Chesapeake | 199,184 |
| Richmond | 197,790 |
| Newport News | 180,150 |
| Hampton | 146,437 |
| Alexandria | 128,283 |
| Portsmouth | 100,565 |
| Roanoke | 94,911 |
| Lynchburg | 65,269 |

According to the 2000 census, Virginia's population is made up of 72.3% whites, 19.6% African-Americans, 3.7% Asians, 0.3% Native Americans, and 0.1% Native Hawaiians and other Pacific Islanders, with the remaining 4% reporting either a mixed heritage or not reporting race. Of these, 4.7% reported a Hispanic background, and 11.1% of Virginians reported speaking a language other than English at home. The centers of the state's metropolitan corridor have the greatest ethnic diversity, with one of the nation's largest Filipino communities in Norfolk as well as large Vietnamese, Korean, and Central American Hispanic communities in Northern Virginia. An increase in the percentage of Hispanic and Asian Virginians is expected in the future, as is true nationwide. The religious practices of Virginians are varied; however, the majority is Protestant, particularly Baptist and Methodist. There are also large Roman Catholic and Jewish congregations in the state. The 2000 census reported that approximately a quarter of the population in Virginia, over 1.7 million, are children under 18, including almost 500,000 ages 4 or younger (3). Education in Virginia is compulsory between the ages of 5 and 18, with an average of $6491 per student spent by the state for the 1999-2000 school year (national average = $7392). The student to teacher ratio was 12.5:1 compared to the national average of 16:1. According to the 2000

census, 81.5% of Virginians age 25 and older had a high school diploma, while 29.5% had a bachelor's degree or higher.

### The Health of Virginians

Virginia generally fares well on various health parameters compared to other southern states, with an average to slightly above average ranking compared to national standards. There remain significant gaps, however, and certainly much room for improvement. The overall life expectancy for Virginians in 2001 was 77.2 years, the same as the national average. There were significant differences in life expectancy between races, however, with a life expectancy of 78.1 years for whites versus 72.8 for African-Americans in Virginia (8). In its *Healthy Virginia Communities* 2000 report, VDH indicated some improvements in several key health risk indicators focused on three goals: improving pregnancy outcomes, decreasing the burden of chronic disease, and protecting Virginians from communicable diseases and environmental health hazards. Notable exceptions included declines in recent Pap tests and decreased physical activity, as well as numerous parameters in which no significant change was noted over the three-year study period (9).

Perhaps influenced by its history of tobacco farming and cigarette production, Virginia has long had a higher percentage of smokers than many other states, although rates in the state as well as nationally continue to decline (10). As the number of smokers declines, the incidence of lung cancer and other tobacco-related disease will decrease, however, these disorders will not disappear, because some cases are largely due to genetic factors. This illustrates an important medical principle: while currently a high percentage of lung cancer is due to the effects of smoking, once that environmental toxin is reduced or eliminated, the remaining cases of disease will include a high proportion with a hereditary origin. Thus the disease will have become one that is largely genetic, although currently it is thought of as being mainly environmental in origin (11).

The leading causes of death in the United States-heart disease, cancer, and stroke-have remained virtually unchanged for decades. The top ten causes of death in Virginia in 1998 as reported by the Virginia Center for Health Statistics are shown in Table 3-3. These largely reflect national data, although septicemia was not among the top ten causes of death nationally, while chronic liver disease and cirrhosis was in the top ten (8, 9). With the exception of unintentional injury, all of these disease categories are influenced by genetic factors.

**Table 3-3. Ten Leading Causes of Death in Virginia, 1998**

| |
|---|
| 1. Diseases of the heart |
| 2. Malignant neoplasms |
| 3. Cerebrovascular diseases |
| 4. Chronic obstructive pulmonary diseases |
| 5. Unintentional injury |
| 6. Pneumonia and influenza |
| 7. Diabetes mellitus |
| 8. Suicide |
| 9. Septicemia |
| 10. Nephritis, nephrotic syndrome and nephrosis |

The health status of minorities in Virginia is of particular concern, becausse minorities, particularly African Americans, tend to lag behind in many categories, a difference largely thought to be caused by

socioeconomic differences. A 1999 VDH report highlighted the disparities affecting the health of Virginia's minorities:

- Hispanic women had the highest birth rate (24.6/1000 population).
- Two-thirds of black infants and one-third of Hispanic infants were born to unmarried mothers.
- Education levels among Hispanic mothers were much lower than whites and Asian/Pacific Islanders.
- Low birth weight among blacks is approximately twice that of other racial groups.
- Less than 75% of black and Hispanic women obtain prenatal care in the first trimester.
- Fetal death and infant mortality are highest among blacks.
- Teen pregnancy rates are highest in black and Hispanic populations (12).

Such disparities must be considered as statewide health programs, including genetic services, are continued and developed further.

Infant mortality rates are another indicator commonly used to describe the health status of a population. The rates have generally been declining in Virginia and the United States overall, with a 90% decrease in infant mortality and a 99% decrease in maternal mortality since 1900. These improvements are largely the result of better hygiene and nutrition, the availability of antibiotics, improved access to health care, and technological advances in obstetric and neonatal medicine (13). Unfortunately, infant mortality rates in the United States remain significantly higher than many other industrialized nations, where rates as low as 2.4 per 1000 were reported by Sweden and Iceland in 2000 (14). The 2001 infant mortality rate in Virginia was 6.95 per 1000. Table 3-4 shows infant mortality rates by race and Health Services Area (HSA), clearly indicating great disparity between races and localities (15). The map of Virginia on the next page indicates the boundaries of the five HSAs (16). A 2001 report on child deaths in Virginia by the Virginia State Child Fatality Review Team noted that 80% of deaths to Virginia children aged 0-17 were deaths of infants. The report also indicated that 68% of infant mortality resulted from congenital anomalies or conditions originating in the perinatal period, many of which are genetic in origin, while 11% were attributed to Sudden Infant Death Syndrome (SIDS) (15).

**Table 3-4. Virginia 2001 Infant Mortality Rates per 1000 Live Births**

| | |
|---|---|
| Statewide average | 6.95 |
| White | 4.28 |
| Black | 14.77 |
| Other races | 8.08 |
| Central HSA | 8.85 |
| Eastern HSA | 9.68 |
| Northwest HSA | 5.73 |
| Northern HSA | 4.35 |
| Southwest HSA | 6.86 |

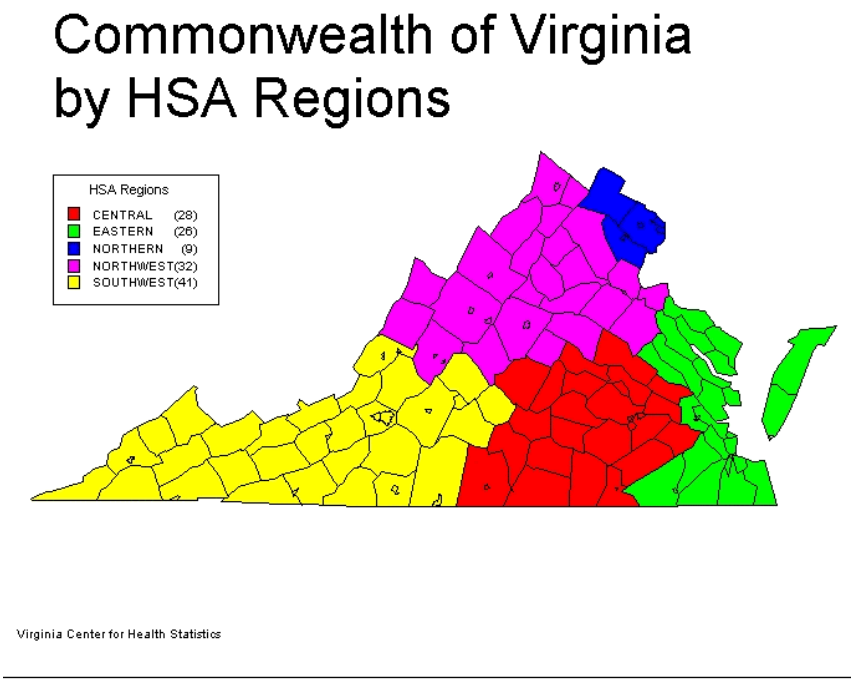
The Annie E. Casey Foundation funds a project known as *Kids Count*, which is focused on tracking various indicators of the educational, social, economic, and physical well-being of children nationally and on the state level. *Kids Count 2003* data on ten key indicators, which is based on statistics from 2000, ranked Virginia at number 14 overall across all states. On individual indicators, Virginia's worst rankings were in the low-birthweight (7.9%; rank 31), infant mortality rate (6.9 per 1000; rank 26) and teen death rate by accident, homicide or suicide (52 per 100,000; rank 21) categories. Much more detailed information, including statistics at the city and county level, is available online at http://www.kidscount.org (2).

Children with special health care needs (CSHCN), many of whom have genetic conditions, are often

poorly served by the current health care system, in Virginia as well as nationally. There are an estimated 9 million CSHCN in the United States, accounting for 13% of all children. Many of these children do not have a medical home or routine source of care, referrals are often difficult, and unmet needs are common (17). A study of Virginia's approximately 240,000 CSHCN identified multiple unmet needs:

- Primary care capacity is strong but not always responsive to the needs of CSHCN.
- Medical specialty capacity is excellent but inconvenient to some parts of the state.
- The special education system is struggling to meet the needs of older CSHCN.
- The system does not support families well, with a lack of information, advocacy and support services.
- Insurance coverage is not equitably and consistently available to families.
- Families in rural areas continue to struggle with excessive travel to medical and support services, despite community-based specialty services in many areas.
- Systems of care for CSHCN are not adequately coordinated or integrated.
- Systems of care for CSHCN do not sufficiently value the input and experience of family members.
- Systems of care for CSHCN are often not culturally competent.
- Many CSHCN remain unidentified because there is no systematic identification strategy (18).

**Figure 3-2. Commonwealth of Virginia by HSA Regions**




The 2000 census reported that over 1 million Virginians aged 5 and above have a disability, amounting to more than 16% of the population. This includes 125,668 (8.1%) of the population aged 5 to 20 (7, 19, 20). Virginia lags behind most of the rest of the country in its efforts toward community placement and community services in particular, with little recent growth and reduced spending in these areas compared to other states. Total spending for community and institutional services for people with mental retardation (MR) and developmental disabilities is also among the lowest in the country (21). The Olmstead Task Force, established in 2002 to assess Virginia's progress toward complying with the Americans with Disabilities Act (ADA), recently reported that there are insufficient community services for disabled Virginians and significant shortfalls in state efforts toward community placement of disabled individuals

who are currently institutionalized because of a lack of state support services. Numerous recommendations were made by the task force in an effort to improve the current situation (22).

### The Contribution of Genetic Disease

Though many genetic disorders are individually rare, there are approximately 15,000 listed in *Online Mendelian Inheritance in Man (OMIM),* a catalogue of identified human genes and genetic conditions, affecting 13 million Americans (23, 24). As information from the HGP and other genetic research projects has grown, it has become obvious that genetic factors contribute to almost all human disease. In particular, the contribution of genetic disease to infant deaths, hospital admissions, MR, and adult chronic disease in the United States is discussed here. Long-term studies of the overall incidence of genetic disease worldwide show no evidence of an increasing frequency of deleterious genes, a concern of some because improved medical care is increasing the survival of individuals with genetic conditions. Social and demographic changes, combined with improved survival of newborns in general, is serving to maintain a constant, or slightly decreasing, incidence of deleterious genes (25). Ongoing efforts toward improved treatment and prevention will undoubtedly continue to decrease the morbidity and mortality associated with these conditions.

Birth defects complicate 3-5% of all births, which translates to approximately 150,000 each year in the U.S., and about 4600 in Virginia (26, 27). A much higher proportion of stillbirths and miscarriages result from congenital anomalies or other genetic conditions. While environmental and nutritional factors contribute to birth defects, many are genetic in origin. Unfortunately, the specific causes of many birth defects are still unknown, although most seem to be due to a combination of factors. The fatality rate among children with birth defects is estimated to be 5.8% by the Virginia Congenital Anomalies Reporting and Education System (VaCARES), a statewide population-based birth defect registry (27). For the 10-year period ending in 1998, VaCARES identified significantly higher birth defect rates for several groups:

- Males (5.5% versus 4.4% in females)
- Blacks (7.1% versus 4.3% in whites)
- Multiple gestations ($\geq$9.2% versus 4.8% in singletons)
- Low birth weight babies (14.2% versus 4.2 % in full birth weight babies)
- Preterm births (10.8% versus 4.2% in fullterm births)
- Babies born to mothers less than 20 years of age (6.5% versus 4.2% for those born to mothers over 20)
- Babies born to mothers who smoked and/or drank alcohol (6.0-8.1% versus 4.8% for non-smokers/drinkers) (27).

An estimated 20-30% of infant deaths in the U.S. are due to birth defects, making them the leading cause of infant death in this country. In addition, some infant deaths attributed to other causes may in fact have an undiagnosed genetic etiology, as demonstrated by a study of SIDS in Virginia, in which at least 1% of SIDS cases had a treatable metabolic genetic disorder (28). Of all newborns, 0.5% have a chromosomal abnormality, however, many more miscarriages and stillbirths are affected by a chromosome disorder (29, 30). The Virginia-specific incidence of several of the most common birth defects as reported by VaCARES is listed in Table 3-5 (27).

**Table 3-5. Selected Common Birth Defects in Virginia**

| Birth Defect Category | Estimated Incidence as a % of Live Births |
|---|---|
| Cardiovascular anomalies | 1.27% |
| Eye anomalies and vision impairment | 0.24% |
| Ear anomalies and hearing impairment | 0.17% |
| Cleft lip and/or palate | 0.13% |
| Down syndrome | 0.09% |
| Hydrocephalus | 0.08% |
| Neural tube defects | 0.06% |

A recent study of admissions to a pediatric hospital in Ohio demonstrated that 34% of children had a clearly genetic disorder, while a total of 71% had an underlying disorder with a significant genetic component, illustrating the major impact of genetic disease on health care in this country (31). Even among adults, an estimated 12% of hospital admissions are for genetic causes (32). For individuals with birth defects, hospital charges for an average stay in 2000 were $32,000, not including physician charges or other outpatient medical services, an indication of the overwhelming financial cost of genetic disorders. Beyond emotional and social costs, the lifetime financial burden of birth defects in the U.S. is estimated at $8 billion (33). A study of visits to a pediatric emergency room in an urban U.S. hospital showed that almost 20% were related to genetic disorders. Of these, over 97% involved complaints or diagnoses that suggested the possibility of an underlying genetic disorder requiring further evaluation and diagnostic work-up, implying that many children with genetic conditions remain undiagnosed until an acute event occurs (34). A high proportion of patients in neonatal intensive care units (NICU) are affected by genetic disorders, including chromosomal abnormalities, Mendelian conditions, single primary developmental anomalies, and unrecognized patterns of malformation. A five-year study of an Arkansas NICU indicated that 23% of patients had a genetic disorder. The study also compared medical records with vital statistic records for cause of death and found discrepancies in 40% of cases, indicating a pattern of under-recognition of genetic disorders by current vital statistics collection methods (35).

Mental retardation (MR), a symptom found in a very heterogeneous group of disorders, affects approximately 3% of the U.S. population (36). An estimated 50% of MR has a genetic basis, while more than 900 genetic syndromes are associated with MR (32, 36). Fragile X syndrome and Down syndrome are among the most common causes of syndromic MR, or disorders in which there are other manifestations in addition to MR. More recently, several different genetic changes have been associated with nonspecific MR, or disorders in which MR is the only primary symptom. In some cases, elucidation of the hereditary basis of disease has resulted in the development of treatments that prevent or minimize the associated MR, such as in PKU and several other disorders commonly included in statewide newborn screening programs. Ever increasing knowledge about the genetic basis of other MR disorders will undoubtedly lead to improved treatment regimens and a reduction in the incidence of MR in the future.

An estimated 15% of cancers result largely from inherited cancer syndromes, while at least 10% of cases of chronic diseases in the U.S. adult population, including heart disease, diabetes, and other disorders, are caused predominantly by genetic factors (37, 38). Many more cases of chronic disease seem to be influenced to a somewhat lesser degree by inherited susceptibilities, although the complete extent of genetic influence on these common diseases is currently impossible to accurately determine. As genetic technologies have improved in the last decade or so, research aimed at detecting the underlying disease susceptibility genes has exploded, including studies of asthma, autoimmune disorders such as rheumatoid arthritis, mental illness, obesity, and even susceptibility to infectious disease. Ultimately, it is hoped that this research will not only lead to a better understanding of the causes and mechanisms of disease but also

to improved treatments and prevention.

The worldwide burden of disease attributable to genetic causes is impossible to estimate, particularly since genetic factors contribute to almost all diseases; however, the World Health Organization (WHO) has estimated the burden attributable to certain hereditary disorders individually. WHO estimates that almost 70% of cases of thalassemia and sickle cell disease worldwide occur in sub-Saharan Africa, while about 95% of cases of cystic fibrosis in Latin America are thought to remain undiagnosed. Approximately 420,000 individuals worldwide are estimated to have hemophilia. About 4 million fetuses and infants each year are born with major congenital malformations. While the avoidable burden of genetic disease cannot currently be quantified, particularly for common chronic diseases influenced by multiple genes, it will probably be substantial even if only a small fraction of the attributable burden is reversed (39).

## 4. GENETIC SYSTEMS OF CARE IN VIRGINIA

Over the past few decades, clinical genetic services have been developed throughout Virginia. These systems of care have gradually grown and changed over time, in response to many factors, including the growing population in many regions, the changing methods of health care management, and the expansion of genetic knowledge, particularly as it affects human disease. No doubt future changes in the genetic systems of care for Virginia will also be necessary.

### Clinical Genetic Service Centers

Several centers providing clinical genetic services are located throughout the state. The largest of these are housed in academic medical centers in Charlottesville, Norfolk, and Richmond, while services in Northern Virginia, with the largest population concentration in the state, are currently in transition. Centers providing limited genetic services are located in a variety of other locations in the state. The map below indicates localities where genetics clinics are held, including outreach clinic sites (1). Table 4-1 indicates recent patient volumes at many of these centers.

**Figure 4-1. Genetic Clinic Locations, Virginia**


Adapted from GeneTests/GeneClinics

Charlottesville is the headquarters for several specialized genetics clinics that are part of the UVA Health System. The Division of Medical Genetics in the Department of Pediatrics provides genetics services to children and adults with a wide variety of genetic conditions and is staffed by three clinical geneticists and three genetic counselors. Services include inpatient and outpatient consultations, diagnostic services, genetic counseling, and management recommendations. Laboratories providing cytogenetic, biochemical, and molecular genetic testing are on-site. The division also staffs several specialty clinics, including the only comprehensive Huntington disease predictive genetic testing clinic in the state. The division has the most extensive system of outreach clinics in Virginia, with clinics of varying frequency held in Abingdon, Roanoke, Lynchburg, and Winchester as well as Beckley, West Virginia (2). Postdoctoral training, including a clinical genetics fellowship, is offered by the program (3). Prenatal genetic services are available through the Prenatal Diagnosis and Treatment Center, part of the Division of Maternal-Fetal Medicine in the Department of Obstetrics and Gynecology of the UVA Health System. These services are provided by two genetic counselors in conjunction with several perinatologists. UVA also has a Cancer Genetics Clinic staffed by one genetic counselor in the Department of Internal Medicine (4). Prenatal genetic services in Charlottesville are also provided by an independent group at Martha Jefferson

Hospital, which is staffed by two genetic counselors.

**Table 4-1. Total Patient Volume Estimates at Selected Clinical Sites**

| | 1998 | 1999 | 2000 | 2001 | 2002 | 2003* |
|---|---|---|---|---|---|---|
| **UVA** | | | | | | |
| Prenatal | 1444 | 1037 | 1405 | 1763 | 1017 | --- |
| General | 329 | 348 | 449 | 350 | 369 | --- |
| Total | 1773 | 1385 | 1854 | 2113 | 1386 | 1066 |
| **VCU** | | | | | | |
| Prenatal | 1169 | 1135 | 1100 | 1100 | 1150 | 967 |
| General | 540 | 585 | 653 | 698 | 775 | --- |
| Cancer | 10 | 15 | 41 | 84 | 81 | --- |
| Total | 1719 | 1735 | 1794 | 1882 | 2006 | 1760 |
| **CHKD** | | | | | | |
| General | 374 | 401 | 393 | 406 | 554 | 1418 |
| **NMCP** | | | | | | |
| Prenatal | 700 | 965 | 1115 | 1045 | --- | --- |
| Cancer | 0 | 3 | 10 | 5 | Not offered | --- |
| **GIVF** | | | | | | |
| Prenatal | 2600 | 2400 | 2200 | 1950 | 1900 | --- |
| Cancer | --- | --- | --- | --- | --- | ---- |

* Calculated data

Richmond is the headquarters for genetic services provided through the Department of Human Genetics on the MCV campus of VCU. Services include inpatient and outpatient consultations, diagnostic services, genetic counseling, and management recommendations. Laboratories providing cytogenetic and molecular genetic testing are on-site. The department provides general genetics services for children and adults at two sites in Richmond as well as one outreach clinic in Fredericksburg. In addition, metabolic outreach clinics are held in Newport News and Fredericksburg. Prenatal genetics clinics are held at five sites throughout the Richmond metropolitan area through contracts with several independent maternal-fetal medicine centers in the area, as well as one site in Fredericksburg. Three full-time and two part-time clinical geneticists and seven genetic counselors and one and one-half cancer genetic counselors staff these genetic clinics. A cancer genetics clinic is held in conjunction with Massey Cancer Center on the MCV campus, staffed by one genetic counselor. At VCU, as at many of the other genetic centers, most of the genetic providers have a variety of other educational and research responsibilities, and thus cannot be devoted to clinical activities on a full-time basis. The Department of Human Genetics at VCU is the most comprehensive genetics center in the state, with a large number of additional faculty members involved in research and educational activities. Educational curricula include programs leading to Masters and Doctor of Philosophy degrees in Human Genetics, as well as a Masters Degree program in Genetic Counseling. Postdoctoral training, including a clinical genetics fellowship, is also offered (5).

The Eastern Virginia Medical Center in Norfolk is the headquarters for several groups providing genetic services, including the Division of Medical Genetics within the Department of Pediatrics of CHKD, The Center for Advanced Fetal Therapy of EVMS, and the Jones Institute for Reproductive Medicine of EVMS. The Division of Medical Genetics is staffed by one and one-half clinical geneticists and two genetic counselors, and provides services for children and adults at their main clinic in Norfolk as well as outreach clinics at three other sites in the Hampton Roads metropolitan area and Nassawadox on the Eastern Shore. Services include inpatient and outpatient consultations, diagnostic services, genetic

counseling for a wide variety of genetic conditions, management recommendations, as well as ongoing management of individuals with selected genetic disorders. A cytogenetic laboratory is on-site (6). The Center for Advanced Fetal Therapy provides a full range of prenatal genetics services, including genetic counseling regarding maternal age issues, abnormal serum or ultrasound screening, family histories of birth defects or other genetic disorders, potential teratogenic exposures, and pregnancy loss issues. The center is staffed by four genetic counselors in conjunction with several perinatologists, with clinics in Norfolk and Newport News (7). The Jones Institute is a comprehensive center of in vitro fertilization (IVF) and related technologies and provides preimplantation genetic diagnosis (PGD) services, although no clinical genetics professionals are on staff (8). No cancer genetics services are offered in the Hampton Roads area.

Other centers providing genetic services in Hampton Roads include Naval Medical Center Portsmouth (NMCP), one of the largest military medical facilities worldwide, which provides tertiary-care services to the large military population in the Hampton Roads area as well as referrals from distant military installations. Prenatal genetic counseling is currently provided by one genetic counselor in conjunction with several perinatologists in the Division of Maternal-Fetal Medicine of the Department of Obstetrics and Gynecology. A clinical geneticist has also recently been added to the staff of the Department of Pediatrics and is initiating a pediatric genetic clinic at NMCP (9). In addition, one clinical geneticist has a solo practice in Virginia Beach, known as the Genetics and Disabilities Diagnostic Care Center, which provides clinical genetic services for children and adults with a variety of genetic conditions (10).

There are currently no comprehensive clinical genetics centers in Northern Virginia, although several sites do provide some genetic services. GIVF, based in Fairfax, provides a full range of prenatal genetic services, as well as being a full-service center for IVF and related technologies, including PGD services. GIVF is staffed by two clinical geneticists and five genetic counselors. Previously, GIVF housed laboratories providing cytogenetic and molecular genetic testing; however, these have recently been acquired by a national commercial laboratory. GIVF also provides genetic counseling and clinical consultation for hereditary cancers but has discontinued its general genetics clinic for children and adults (11). Pediatric genetic services are provided at Inova Fairfax Hospital for Children, which has one clinical geneticist on staff in the Department of Pediatrics (12). Cancer genetic counseling is provided through the Cancer Genetic Counseling and Screening Program at Inova Fairfax Hospital, staffed by one genetic counselor (13). The Children's National Medical Center, based in Washington D.C., holds outreach clinics in Fairfax, Virginia, including pediatric and prenatal genetics clinics, while the Genetic Center of Virginia based in Annandale provides prenatal genetic services as well; unfortunately, staffing and patient volume information for these centers are unknown (1).

A full range of prenatal genetic services is offered at the Carilion Prenatal Diagnostic Center in Roanoke, which is staffed by two genetic counselors in conjunction with several perinatologists (14).

Many Virginians, undoubtedly, obtain genetic clinical care from sites in neighboring states. In particular, the University of Kentucky's Genetics and Dysmorphology Clinic, based in Lexington, Kentucky, holds several outreach clinics near Kentucky's border with southwestern Virginia, a very rural part of the state that is quite distant from genetic centers in Virginia. West Virginia University operates one outreach clinic near the northwestern border of Virginia. In addition, several medical centers in Washington D.C. and southern Maryland have large, comprehensive genetics units that currently must absorb much of the clinical care for the significant population in Northern Virginia, has only limited availability of local genetics services for children and adults (1)

## Genetic Outreach Programs

Underserved populations, largely comprised of groups that are either culturally or geographically removed from the current systems of care, continue to be an issue in clinical genetics. As described in the previous section, most of the large genetics centers do hold outreach clinics in an attempt to reach some of the geographically isolated areas, but there remain unmet needs. Telemedicine has been utilized in other states to facilitate the provision of genetic health care to rural residents, with successful outcomes overall. In Virginia, telemedicine has been used in an ongoing program by UVA neurology providers caring for patients with Huntington disease and has been used intermittently by VCU for cancer genetic counseling as well (15). In Georgia, the Children's Medical Services Program, a Title V program, has developed a network of subspecialty clinics throughout the state for children with special health care needs, staffed by traveling physicians and supplemented by telemedicine clinics. A variety of subspecialists, including genetic providers, provide consultations via this telemedicine network, which allows for more frequent subspecialty appointments as well as travel cost savings. With increasing experience, providers have become more comfortable providing a variety of services remotely, including new patient consultations, and have increased their efficiency with the telemedicine equipment. Surveyed patients and providers have been satisfied with the telemedicine service and support their continuation as a supplementary mode of care delivery for rural populations (16). Maine has one of the largest telemedicine systems in the country, with more than 150 participating facilities, including a mobile telemedicine boat that serves small islands off the Maine coast. Genetic consultations account for approximately 6% of the over 1800 clinical usages annually. Telemedicine in Maine is also used for educational and administrative meetings throughout the state, resulting in time and cost savings for participants and sponsoring organizations. As in Georgia, patients and providers have been quite satisfied with the telemedicine consultations. Reimbursement for telemedicine visits in Maine has improved, and their system is approaching fiscal sustainability (17).

Although the Hispanic population in Virginia is not as large as in many other states, it is a growing minority with significant unmet health care needs. In particular, there are groups in Northern Virginia as well as a large migrant worker population on the Eastern Shore that speak Spanish as their primary language, with limited English skills. The same is true for several other ethnic groups in various regions of the state. For the most part, genetic providers in Virginia have limited resources to communicate with these patients. Although efforts are made to accommodate them and interpreter services are sometimes available, they are often not ideal. In addition, cultural differences of minority groups in the state are often not well addressed within current genetic service models, which have traditionally been based on the majority culture. While genetic centers in other areas of the country have begun providing foreign language training or recruiting genetic professionals that already possess these skills, such proactive efforts have not occurred in Virginia, perhaps because the need has not yet reached some critical level (18, 19). Additional resources will be needed to address growing diversity issues in the future.

## Workforce, Workload, and Manpower Issues

The sufficiency of the clinical genetics workforce in the U.S. has long been an area of inquiry. More than 30 years ago, the National Institute of General Medical Sciences, Congress, and the National Institutes of Health singled out genetics as a field of medicine with great needs for manpower expansion, at a time when it was estimated that less than 25% of families with genetic conditions were offered genetic counseling (20). Despite this, the field remains very small, with little recent workforce growth overall. Following the 2005 board certification exams, which are offered every three years, there were approximately 1178 certified clinical geneticists and 2171 certified genetic counselors; however, these numbers include individuals that may no longer be practicing (21, 22). While the number of genetic

counselors in practice continues to rise, the number of physicians sitting for the clinical genetics boards has gradually decreased over the past decade. In Virginia, there are an estimated 55 clinical geneticists and genetic counselors, although not all are in full-time genetic practice. Kaiser Permanente, a large health care organization with medical facilities in various other areas of the country, recently estimated their internal staffing needs for appropriate genetic care. Extrapolating that data to Virginia's population count from the 2000 census indicates that 26 clinical geneticists and 106 genetic counselors are necessary to meet current population needs in Virginia (23).

Data from a 2002 National Society of Genetic Counselors (NSGC) survey indicates that the majority of genetic counselors in the U.S. and Canada work in prenatal settings. Traditionally, pediatric settings have been the second most common, but by 2002 cancer genetic counseling had increased beyond the static pediatric setting levels. The number of genetic counselors in other areas--particularly other adult disease clinics, commercial genetic laboratories, and clinical research--is also increasing. This trend is likely to continue as our knowledge of the genetic basis of common adult disease grows and is reflected to some extent in Virginia, where a few genetic counselors are involved with cancer genetics clinics, while several work for genetic laboratories and are not directly involved in patient care. Most genetic counselors report that a majority of their time is spent on clinical care, with teaching and research responsibilities also being significant (24). Although most pediatric genetic care is provided by teams consisting of genetic counselors working directly with clinical geneticists, for the most part, genetic counselors working in other areas—including prenatal and cancer settings—provide genetic care more independently, often as the genetic expert on a team that includes physicians from non-genetic specialties. Over 90% of genetic counselors are Caucasian females, which compounds the gaps regarding minority language and cultural issues. Most genetic counselors in Virginia and the country overall work in urban areas with large populations, although only 43% currently report a university medical center as their primary work setting. The geographic distribution has become more widespread in some areas of the country recently as the total number of genetic counselors in practice has increased. A 2000 genetic counselor workforce study concluded that the current level of genetic counselor training should be maintained, with monitoring of the growth in clinical genetic testing and demand for genetic counselors' services. Expansion was not thought to be feasible currently given poor financial support for clinical genetics, including training programs and genetic counselors in practice (25).

Although there are few in Virginia, genetic nurse specialists are part of the clinical genetics workforce in some areas of the country, often filling roles similar to those of genetic counselors. The International Society of Nurses in Genetics (ISONG) is an active professional organization promoting an expansion of genetics training in core nursing curricula and sponsors programs that offer advanced training in human genetics for nurses (25).

As part of the Future of Pediatric Education II (FOPE II) Project, board certified clinical geneticists were surveyed regarding workload and practice issues. As is the case for genetic counselors, the majority of clinical geneticists in the U.S. are white, although there are slightly more males than females, and the percentage of represented minorities is somewhat higher. In addition to genetics training, most of the survey respondents had specialty training in general pediatrics, while a few had obstetrics and gynecology, maternal-fetal medicine, internal medicine, or other specialty training. Over 80% of respondents work in urban settings, largely in academic medical centers, with most referrals coming from pediatricians, although referrals from adult subspecialists represent the largest referral growth area. Geneticists' income, among the lowest in U.S. medical schools, largely comes from salaries, although some reported income from fee-for-service or prepaid capitated sources (26, 27). With an average workweek of 57 hours combined with relatively low incomes, clinical genetics is not likely to be attractive to future physicians without significant practice changes, as a recent survey of Virginia medical

students indicated that time commitment in practice and expected salary are the most important influences on career choice, along with academic interest (27, 28).

Clinical genetic care includes a large, labor-intensive cognitive component, with an emphasis on preventive health care. The nature of genetic conditions results in a significant amount of time spent researching rare disorders that are part of the differential diagnosis, reviewing extensive medical records, explaining complex genetic concepts to patients and families, and arranging a variety of molecular tests. Geneticists and genetic counselors have also traditionally sent a written summary to referring physicians as well as patients, which requires additional time. Research on workload in the 1980s showed that the average amount of time devoted to a single new patient general genetics clinic visit, including preparation and research done before and after the visit, was 7.1 hours, while time for a follow-up visit was 4.0 hours, with clinical practice income providing only 37% of personnel costs (29). By the time the FOPE II survey was performed in the late 1990s, reported time spent for average patient visits had dropped to 3.1 hours for new patients and 1.4 hours for follow-up visits; however, the recent numbers reflect the time spent by the geneticist only, not genetic counselors or other support personnel, as the previous estimates had. On average, geneticists reported spending approximately half of their time in direct patient care, seeing an average of seven new and six follow-up patients per week, with the remainder of their time divided between basic research, teaching, clinical research, and clinical genetic laboratory responsibilities (27). Genetic counselors report an average of eight new and three follow-up patients per week, although this varies tremendously depending on work setting (24). Geneticists reported that two-thirds of their patient volume was comprised of children, with the remainder being prenatal or adult patients, while the majority of patient care was provided in outpatient settings, although geneticists are frequently involved with multispecialty clinics and inpatient care as well. Poor reimbursement for clinical care by third-party payers continues to be a major problem for genetic professionals, exacerbated by the fact that genetic counselors generally cannot directly bill insurers for their services and income from in-house clinical genetic laboratories, a significant source of support for clinical services previously, has been drastically reduced as the result of contracts between insurers and commercial laboratories. (27).

A major question regarding future workforce planning for geneticists revolves around what role they will play in the overall health care scheme in the future. If clinical geneticists continue to provide comprehensive genetic care for an ever-increasing patient population as medical technology expands, then a much larger workforce is needed; however, many experts believe a more likely scenario is one in which geneticists play a larger role in educating PCPs, who will then incorporate more extensive genetic care into their daily practice. With some reports of general practitioners spending an average of 7 minutes per patient, appropriate genetic care would obviously require significant changes in generalist practice patterns for the latter scenario to be acceptable (30). The future role of genetic counselors, and thus workforce needs, is also uncertain, although recent trends suggest that genetic counselors are increasingly working directly with other non-genetic medical specialists as part of health care delivery teams (27). Although much remains uncertain, it seems clear that clinical geneticists and genetic counselors will increasingly be involved in three main roles:

- Providing genetic counseling and testing for complex genetic conditions;
- Providing genetic training and education for other health professionals; and
- Providing specialized genetic services (31).

## Virginia's Genetic Legislation

A variety of state and federal legislation is relevant to the provision of genetic health care services. In Virginia, several laws address issues of genetic privacy and nondiscrimination, while other legislation requires a variety of genetic programs within the state's public health system. Genetic issues related to

assisted reproduction, surrogacy, establishment of paternity, and court evidence are also discussed within the *Code of Virginia* (32).

Laws in all states restrict access to medical records, including genetic information; however, several states, including Virginia, have laws specifically pertaining to the privacy of genetic information. Virginia's genetic privacy law prohibits the disclosure of genetic information without informed consent. Some other states require informed consent for a third party to perform or require a genetic test, or to obtain or access genetic information. A few states define genetic information as personal property and some have established specific penalties, civil or criminal, for violating genetic privacy laws. Genetic nondiscrimination in health insurance is addressed at the federal level by the Health Insurance Portability and Accountability Act of 1996 (HIPAA), which prohibits health insurance discrimination based on any "health status-related factor," including genetic information, for group health plans, which are usually those with more than 50 individuals. Various state laws provide stricter prohibitions on genetic discrimination in health insurance. In Virginia, legislation applying to both individual and group insurance policies prohibits the establishment of rules for eligibility based on genetic information, prohibits the use of genetic information for risk selection or risk classification, and prohibits disclosure of genetic information without informed consent. Genetic information in this case includes genetic test results, genetic test results of family members, and inherited characteristics. Laws regarding employer use of genetic information were enacted in several states in the 1970s and 1980s in response to employer discrimination against applicants with sickle cell trait. Virginia's law dates from 2002 and prohibits employer discrimination in hiring, firing, or determining the conditions or privileges of employment based on genetic test results or inherited characteristics. Employers in Virginia are also prohibited from requesting or requiring genetic information or genetic testing and from performing genetic tests on employees. On the federal level, the Equal Employment Opportunity Commission provided an interpretation of "disability" in the ADA that included genetic predisposition to disease, but there is some debate about whether the Supreme Court would accept that interpretation (33). In February 2000, President Clinton banned genetic discrimination in the federal workforce; however, despite the introduction of several bills specifically addressing genetic discrimination, none has been passed by Congress thus far (34). Some concern about genetic discrimination remains, however, as existing laws provide little protection against discrimination in life, disability, or long-term care insurance (35).

State legislation also requires several genetic programs within VDH. The organizational structure of genetic programs within VDH is shown in Figure 4-1, while the programs themselves are further described below.

Programs relevant to genetics within the Division of Women's and Infants' Health include a variety of Perinatal Health and Prevention Programs as well as the *Virginia Sickle Cell Awareness Program* (VASCAP). The aim of VASCAP is to improve the health care system for individuals with sickle cell disease in Virginia. Perinatal programs include the *Regional Perinatal Councils* (RPC)*, BabyCare*, *Resource Mothers,* and *Virginia Healthy Start Initiative,* all of which provide services that may reduce the incidence and morbidity of birth defects. There are seven RPCs in the state, charged with improving the infrastructure through which perinatal health care is provided in Virginia. *BabyCare* provides intensive case management services for pregnant women and infants at risk of poor outcomes who are also Medicaid recipients. Services include risk assessment, coordination of services, follow up, and monitoring, as well as education and counseling as needed. *Resource Mothers* is a program that uses lay community mentors to provide intensive home visiting services to pregnant teens through the infant's first birthday, while Virginia Healthy Start Initiative is a federal program that elaborates on the activities of Resource Mothers in four communities with particularly high infant mortality and morbidity rates in Virginia (36)

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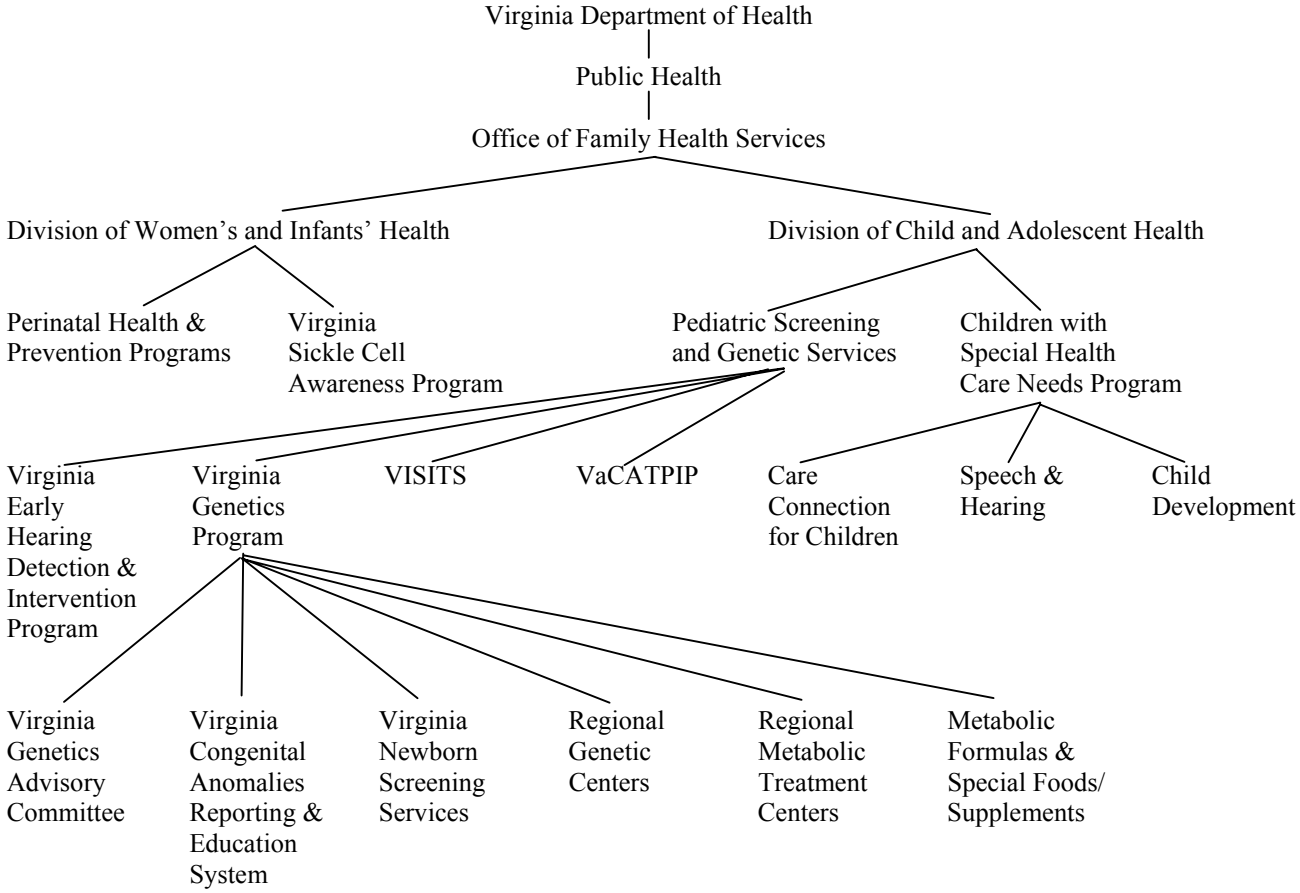
**Figure 4-2. Organizational Structure of Genetic Activities in VDH**




PSGS is charged with improving the health of children and adolescents by preventing birth defects and developmental disabilities, promoting optimal child development, and promoting health and wellness among children and adolescents with disabilities (40). Within PSGS, the goal of the *Virginia Early Hearing Detection and Intervention Program* is to identify congenital hearing loss in children by 3 months of age and enroll them in appropriate early intervention services by 6 months of age. To facilitate this goal, as of July 1, 2000, all Virginia hospitals with newborn nurseries must provide universal hearing screening of newborns before discharge (41). The *Virginia Infant Screening and Infant Tracking System (VISITS)* project involves the development of a Web-based integrated database system that tracks and manages data collected from four programs: *Virginia Early Hearing Detection and Intervention Program, Virginia Newborn Screening Services, VaCARES,* and *At-Risk Screening Program,* a program within CSHCN for children with developmental disabilities (42). The goals of the *Virginia Congenital Anomalies Tracking and Prevention Improvement Project* (*VaCATPIP)* project are to improve and enhance Virginia's population-based birth defect surveillance, prevention, and service program, *VaCARES,* and to integrate surveillance data with public health programs (43). The *Virginia Genetics Program* is responsible for reducing unnecessary morbidity from potential or existing genetic conditions by assuring access to the appropriate education, testing, counseling, and treatment for Virginians, and is coordinated by a nurse manager who also serves as the state genetics coordinator (44). Within this program, the *Virginia Genetics Advisory Committee* (*VaGAC)* provides guidance on planning, implementing, and evaluating genetic services in Virginia, with current goals centered around the development of a state clinical genetics plan, evaluating the implications of tandem mass spectrometry

technology for metabolic disease screening of newborns in Virginia, and involving parents in *VaGAC* activities (45). The *Virginia Genetics Program* also provides some financial support for four *Regional Genetic Centers* that provide genetic testing, counseling, and education in Virginia. Currently, supported programs include the Department of Human Genetics at VCU in Richmond, the Division of Medical Genetics at CHKD in Norfolk, and the Division of Medical Genetics at UVA in Charlottesville, as well as prenatal genetic services at GIVF in Fairfax. *VaCARES*, established by state legislation in 1986, is a birth registry of children with birth defects under the age of 2 that is charged with collecting relevant epidemiological data to evaluate possible causes of congenital anomalies, improving diagnosis and treatment, and educating families about available resources. To facilitate complete surveillance, the *Code of Virginia* mandates universal reporting of birth defects in the state, currently accomplished by collecting information from birth certificates, death certificates, hospital discharge reports, and the *Virginia Newborn Screening Services* database (44, 46). The Department of Defense has also recently initiated a birth defects registry of all military beneficiaries, which includes a significant proportion of Virginia's population (47). *Virginia Newborn Screening Services* provides newborn testing for certain several heritable disorders and genetic diseases that, in the absence of early treatment, result in serious complications, including MR, permanent disability, or death, and also provide referral for appropriate counseling and treatment of affected infants. Infants under 6 months of age who are born in Virginia are screened for the following conditions, which are identified through newborn dried-blood-spot screening tests.

1. Argininosuccinic aciduria (ASA)*
2. Beta-Ketothiolase Deficiency (ßKT)*
3. Biotinidase deficiency (BIOT)
4. Carnitine uptake deficiency (CUD)*
5. Citrullinemia (CIT)*
6. Congenital adrenal hyperplasia (CAH)
7. Congenital hypothyroidism (CH)
8. Cystic fibrosis (CF)*
9. Galactosemia (GALT)
10. Glutaric acidemia type I (GA I)*
11. Hemoglobin Sickle/Beta-thalassemia (HB SßTh)
12. Hemoglobin Sickle/C disease (HB S/C)
13. Homocystinuria (HCY)
14. Isovaleric acidemia (IVA)*
15. Long chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)*
16. Maple syrup urine disease (MSUD)
17. Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
18. Methylmalonic acidemia (mutase deficiency) (MUT)*
19. Methylmalonic acidemia (Cbl A,B)*
20. Multiple carboxylase deficiency (MCD)*
21. Phenylketonuria (PKU)
22. Propionic acidemia (PROP)*
23. Sickle cell anemia (Hb SS disease) (Hb SS)
24. Tyrosinemia type 1 (TFP)*
25. Trifunctional protein deficiency (TFP)*
26. Very long-chain acyl-CoA dehydrogenase deficiency (VLAD)*
27. 3-hydroxy 3-methyl glutaric aciduria (HMG)*
28. 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)*

*As of March 1, 2006

The *Code of Virginia* mandates the testing of all infants in the state unless a parent or guardian objects on religious grounds. Newborn screening within Virginia will be expanding in March of 2006 through the use of tandem mass spectrometry to include additional conditions for which screening is recommended by ACMG and MOD. The *Virginia Genetics Program* also includes *Metabolic Treatment Services,* which include provision of metabolic formulas and reimbursement for low-protein food/special supplements for certain populations. Participating metabolic treatment centers are currently located within the genetic centers at UVA, VCU, and CHKD (44).

For the results of the HGP and other genetic research endeavors to be used to their fullest potential, the role of genetics in public health systems needs to expand significantly. The translation of the new genetic information into the prevention of disease and improvement of health requires the application of core public health principles, including assessment, policy development, and assurance. Public health systems in Virginia and elsewhere are being tasked with new and expanded responsibilities, including monitoring the genetic health of the population; informing, educating, and empowering the population to achieve its best genetic health status; mobilizing community partnerships to accomplish these goals; and developing genetic health policies (48). While Virginia has a history of undertaking these responsibilities in some arenas, their application has expanded rapidly and will continue to do so in the future. Currently, there are no programs within VDH that directly address genetic issues related to adult disease, one of the most rapidly growing areas of genetic medicine. That and other challenges will need to be addressed more thoroughly in anticipation of future needs in clinical genetics in Virginia.

## A Look at Other Systems

Many organizations at the national level provide information, education, and guidance relevant to genetics. Some of these are listed in Table 4-2 along with contact information.

The Newborn Screening Task Force, a federal initiative supported by the American Academy of Pediatrics (AAP) and MCHB, published recommendations regarding newborn screening in August 2000. The report, *Newborn Screening: A Blueprint for the Future,* calls for states and the federal government to work together to create a national model standardizing the structure and function of newborn screening programs. Currently, programs vary significantly between states. Specifically, the report recommends the use of a comprehensive systems approach, following accepted national guidelines, coordination of infant programs and data, piloting of new tests and technologies before adopting major policy changes and new mandates, monitoring and evaluating program performance, involving and informing families, establishing a state advisory group that has a diverse representation, setting state-level policies for the use and storage of residual newborn screening blood samples, and assuring adequate financing (49). In addition, the National Newborn Screening and Genetic Resource Center (NNSGRC) has conducted site visits of newborn screening programs in several states, resulting in specific guidance for systems improvement, an avenue that may be appropriate for Virginia as well.

**Table 4-2. Genetic Resource Organizations**

| | |
|---|---|
| American College of Medical Genetics | http://www.acmg.net |
| Genetics and Educational Materials (GEM) Database. | http://www.gemdatabase.org/GEMDatabase/index.asp |
| GeneTests/GeneClinics | http://www.genetests.org |
| Genetic Alliance | http://www.geneticalliance.org |
| Genetics Education Center | http://www.kumc.edu/gec/ |
| March of Dimes | http://www.modimes.org |
| Health Resources and Services Administration (HRSA) | http://www.hrsa.gov/ |
| HRSA, MCHB, Division of Services for Children with Specials Health Needs, Genetic Services Branch | ftp://ftp.hrsa.gov/mchb/factsheets/dschsn.pdf |
| HRSA, MCHB, Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children | http://mchb.hrsa.gov/programs/genetics/committee/ |
| National Birth Defect Prevention Network | http://www.nbdpn.org |
| National Center for Birth Defects and Developmental Disabilities | http://www.cdc.gov/ncbddd |
| National Conference of State Legislatures | http://www.ncsl.org/programs/health/genetics.htm |
| National Human Genome Research Institute | http://genome.gov/ |
| National Newborn Screening and Genetic Resource Center | http://genes-r-us.uthscsa.edu/index.htm |
| National Organization of Rare Diseases | http://www.rarediseases.org |
| National Society of Genetic Counselors | http://www.nsgc.orghttp://www.nsgc.org/ |
| Office of Genomics and Disease Prevention | http://www.cdc.gov/genomics/default.htm |
| Online Mendelian Inheritance in Man | http://www3.ncbi.nlm.nih.gov/Omim |

A variety of systems of genetic care have been proposed or tried in other states and countries. One of the efforts of CORN, a cooperative organization of 10 U.S. regional genetic services networks established in the 1980s, was to improve the collection of genetic services data nationwide as a basis for documenting utilization of genetic programs. These efforts included the collection of data on prenatal genetic services in a manner similar to methods already used by state departments of public health for the collection of information about newborn screening and pediatric genetic services (50). In Virginia, no information is available about prenatal genetic services beyond the limited data discussed in this report. Some states have analyzed the data collected by their health departments regarding the services provided for genetic patients, including pediatric and prenatal evaluation as well as laboratory procedures, to facilitate program assessment, targeted development of services to underserved and rural areas, coordination of care with other state programs, and evaluation of progress toward national and state health goals as they relate to genetic disease (51). In Virginia, data is collected from the main genetic centers that receive financial support from VDH through Title V block grants. This shared data has begun to be linked to statewide planning.

Genetic support groups provide a critical adjunct role for individuals with genetic conditions and their families. The *Genetic Alliance,* formerly known as the *Alliance of Genetic Support Groups,* serves as a national forum for genetic voluntary organizations. Major priorities of the organization are to develop genetic support groups as needed and to facilitate the effectiveness and efficiency of existing groups. The *Genetic Alliance* also facilitates partnerships between consumers and professionals, and provides an

online database of genetic voluntary organizations (52).

With the exception of newborn screening and early hearing detection, genetic services in Virginia and other states are largely passive, rather than active, in that the services respond to referrals of individuals or families affected by or at risk for a genetic condition. Historically, some clinical geneticists have provided ongoing care for patients with genetic conditions, while the majority have remained primarily diagnosticians with patient management provided by PCPs or other non-genetic specialists. Management styles vary somewhat among practices throughout Virginia, but follow-up and ongoing care is growing at all centers. Genetics centers in the United Kingdom (U.K.) have evaluated a system of proactive genetic services based on preventive medicine models using a regional genetic family register service. Under this system, systematic follow up of patients as well as proactive contact and genetic counseling of relatives is provided, with permission to contact relatives first obtained from probands (the patient first seen in the family). On an annual basis, patient charts are reviewed and follow up visits are offered. This genetic register approach was found to be highly satisfactory to patients, although patients' knowledge retention was not significantly better than under the traditional system. Patients valued the ongoing interaction and support from the genetic providers, with a large proportion of patients and PCPs indicating that the genetic service should be at least partly responsible for contacting at-risk family members (53).

European genetic services have undertaken a major evaluation of their systems of care, with three main targets, including the Confidential Enquiry into Counseling for Genetic Disorders (CEGEN), which systematically audits the provision of genetic information and referral by non-geneticists in the UK. The other goals of the evaluation process are to document the current state of genetic services in 31 European countries and to investigate the interaction between genetic centers and referring providers in 7 of those countries (54). CEGEN uses an approach that applies principles of adverse event evaluation to the provision of genetic information and referral in routine medical practice. In this study, medical records were examined for written documentation of appropriate information regarding genetic tests or interventions provided to patients known to be at risk that had later had a child with a genetic condition or were diagnosed with a hereditary disorder themselves. The authors contend that these genetic management issues should be subject to QA measures as are other adverse events, including maternal and perioperative deaths (55).

The Michigan Department of Health coordinates and funds a statewide program of genetic services that utilizes five genetic counselor coordinators, each responsible for a region of the state. The coordinator in each region serves as a liaison between the comprehensive genetic service centers and the local communities, with responsibilities including coordination of genetic outreach clinics; genetic education of health professionals, social workers, parent groups, teachers, and others; and monitoring of the newborn screening system in local hospitals. The five regional coordinators have facilitated identification of needs and active assessment of other genetic public health programs, and have vastly improved education of providers at all levels throughout the state (56). The Alberta, Canada Hereditary Disease Program utilizes a similar model, with a community health nurse trained in genetics working within almost every health unit and providing coordination of care for patients requiring genetic services at one of the two regional genetic centers in Alberta. The Alberta program also emphasizes education of providers and the public as a major function of the program (57).

The Wisconsin Stillbirth Service Program (WiSSP) was established as a model for the investigation of the causes of stillbirth. Few areas have programs of consistent, comprehensive etiologic investigation of intrauterine death, although such an assessment can provide critical information for reproductive decision-making and future pregnancy management. WiSSP provides outreach education to all birthing centers in the state, specific protocols for use with all stillbirths, and a community-based system of service

provision. Stillbirth evaluations are performed at the participating hospital, each of which has an in-house contact for coordination of local efforts. Results are sent to the comprehensive genetic center, which allows for expert assessment and statewide data collection. Interpretive letters are sent to referring providers and the family, if requested, and counseling is subsequently provided by the patient's primary provider or WiSSP staff. Results of the program indicate that approximately 40% of stillbirths have an identifiable etiology, often genetic in origin (58). In Virginia, stillbirth assessment varies regionally and is not obtained on a consistent basis, although fetal morbidity reviews may include input from clinical geneticists.

## Evolving Healthcare Needs in Genetics

Although no one has a crystal ball, many have provided insight regarding the potential future of genetic services and the ways in which genetic information will be incorporated into the practice of medicine. Genetic providers and the practice of clinical genetics must continue to evolve to keep up with burgeoning technological advances, and changing clinical practices in response to an increasingly competitive health care marketplace. Advances in information technology, including electronic healthcare records, will play a significant role in the development of new systems of care and the management of complex disease (59, 60). Hopefully, the role of genetic testing and genetic services will continue to move toward a prevention model, as an improved understanding of disease processes allows for the development of preventive therapies. Although pressures to quickly implement new medical technologies will likely grow, continued consideration of the ethical, legal, and social ramifications must remain a prime focus (61).

The classic domain of clinical genetics involves relatively rare single gene and chromosomal disorders, conditions that are not well known to most medical providers, as well as birth defects and developmental disabilities. Undoubtedly, genetic providers will continue to play a major role in the diagnosis, counseling, and, in some cases, management of individuals with these conditions. In the future, given the expectation of a significant expansion in treatment options, genetic providers may become increasingly involved with the ongoing management of these patients. Growth in the area of newborn screening through the incorporation of tandem mass spectrometry technology as well as carrier testing is also likely to increase demand for genetic services. A second domain of the genetic contribution to medical practice, one that is becoming more widespread, involves the subset of patients with common disorders for which a single gene change is a major risk factor for disease. The explosion of clinical cancer genetics in the last 5-10 years is a prime example of this. The development of specialized cancer genetic clinics through direct collaboration between oncologists and genetic specialists provides a model that could be adapted to other diseases as their genetic basis is elucidated. Recognizing this subset of patients within the population affected by these common disorders will require further education of PCPs and specialists, likely to be led by genetic providers and facilitated by the development of tools suited for the purpose. Genetic providers may also need further education about the natural history and current management protocols for these common disorders. Public health expertise is likely to be needed to deal with the possibility of wide-scale screening for these disorders, particularly once treatment interventions targeted toward the hereditary forms of the disorders are developed. A third domain, expected to grow as the data generated by the HGP is analyzed and applied to clinical medicine, involves the elucidation of multiple small genetic contributions that combine with environmental and lifestyle factors to produce common disease. It is expected that testing for the genetic variants involved will facilitate individual risk assessment allowing for targeted treatment and education about lifestyle modification. Future tests for genetic variants will probably also be used to identify subcategories of disease that respond to specific treatments (62, 63). While it is unlikely that genetic providers will frequently provide a direct role in the care of these patients, a significant role in the development of the testing systems, interpretation of

complex test results, and policy development is anticipated (64). To facilitate an evolution of genetic care that is as seamless as possible, the systems involved in Virginia and elsewhere must make concrete plans for new and expanded roles in risk assessment, diagnosis, management, counseling, and test development.

# 5. GENETIC PROVIDER SURVEYS: KEY INSIGHTS, IDENTIFIED NEEDS, AND PERCEIVED BARRIERS

## Overview

A thorough assessment of genetic service needs in Virginia requires input from many stakeholders, including genetic providers, non-genetic healthcare providers, and consumers. Toward this end, a comprehensive survey addressing a variety of issues relevant to genetic services was developed with the assistance of the VaGAC and VDH and distributed to clinical genetic providers throughout the state as part of the needs assessment process. The results provide critical insight into current practices and gaps in genetic health services and education that was not previously available. The survey, which is included as Appendix A, contains questions that address practice patterns and service provision, as well as providers' perceptions of a variety of factors, including the quality and accessibility of genetic services in Virginia, availability of resources, reimbursement issues, legal and ethical issues, the role of state organizations, and educational needs and practices.

Following approval by the VCU Institutional Review Board (IRB), the survey was distributed by electronic and/or postal mail to 65 clinical genetic providers in Virginia. The sample population included all genetic counselors, clinical geneticists, and clinical cytogenetic laboratory directors currently practicing in Virginia as identified through the membership directories of several national genetic professional organizations. One survey was returned due to an incorrect address. Nineteen completed surveys were received, for a response rate of approximately 30%. While it is possible that the views of respondents and non-respondents do not coincide, demographic criteria suggest that the respondents do closely mirror the sample population overall. Of respondents, 11% were male and 89% were female, which exactly reflects the gender ratio of the total sample population. In addition, the professional degrees of the respondents closely reflect those of the sample population, with 68% of respondents having a master's degree, compared to 62% of the total sample population, and 21% of respondents having a medical degree, compared to 25% of the sample population. The remainder is largely made up of individuals with a Ph.D. degree, most of whom do not provide direct clinical services. There were multiple respondents from each of the geographic areas containing large genetic centers, including Charlottesville, Hampton Roads, Northern Virginia, and Richmond, suggesting that results are broadly reflective of genetic providers throughout the state. Further information about non-respondents is not available. All respondents were of Caucasian background, reflecting the relative lack of ethnic diversity in the field overall. Years of practice in the genetics profession varied greatly. Although half of respondents reported less than 5 years of experience, the remainder reported a range from 5-10 years to more than 20 years of experience. A variable level of professional involvement is also reflected by the respondents, with 32% currently working part-time. While 63% of respondents primarily work in academic medical centers, others work in non-academic hospitals, medical practices housing single or multiple specialties, or commercial laboratories.

The survey results identified several common themes regarding the successes of clinical genetic services in Virginia as well as gaps and barriers impeding the provision of genetic care in the state. Virginia's genetic providers consistently lauded three areas: (1) the genetic workforce and services available in the state, including excellent advocacy, support, and education for patients with genetic conditions; (2) the quality of genetic population screening programs such as newborn screening; and (3) the tradition of outreach services to underserved areas of the state. Key gaps identified by Virginia's genetic providers revolve around three issues: funding, awareness, and access. By far the most frequently identified barriers to the provision of appropriate genetic care for Virginians were limited funding, restricted insurance

coverage, and inadequate reimbursement for clinical genetic services and genetic testing. Inadequate knowledge of genetics and awareness of genetic services among healthcare providers and the public was also cited as a significant barrier, as was the restricted geographic availability of services and the limited number of genetic providers, particularly clinical geneticists.

### Genetic Services in Virginia

As described in the previous chapter, several types of clinical settings are found throughout Virginia, and the activities of the survey respondents reflect that variety. In addition, many respondents are involved with more than one type of clinic setting. Slightly more than half of respondents see genetic patients in prenatal settings and 32% work in pediatric genetic clinics. While 21% see patients in general genetic clinics, slightly fewer work in cancer genetics or multidisciplinary settings. The majority of respondents provide multiple types of services, most commonly individual and family genetic counseling and specimen collection for genetic testing. Approximately half of respondents report providing more extensive services, including medical management of genetic conditions, nutritional metabolic management, support services, inpatient consultation, genetic autopsy, and care coordination. One survey question attempted to measure the amount of time spent on various work activities; however, cautious interpretation is necessary since the results include respondents that work part-time. The results do suggest that respondents are involved in diverse activities on a regular basis, including patient care and related administrative activities, teaching, research, study, and non-patient-related administration.

Among respondents who see patients, the yearly patient volume varies considerably, reflecting the fact that some respondents are almost exclusively involved in patient care, while others are more involved in teaching, research, and other activities. In prenatal genetics, individual respondents reported seeing up to 775 new patients per year, while in general/pediatric genetic clinics, respondents reported seeing up to 500 new patients per year. Much lower patient volumes were reported for metabolic and cancer genetics, although volumes for cancer genetic clinics have increased in recent years, a trend that is likely to continue in the future. Institutional patient volumes for the last five years are shown in the previous chapter in Table 4-1. In some areas of the state, an inadequate genetic workforce is suggested by how far ahead genetic clinics are booked, although in all cases, providers strive to triage patients so that critical patients are seen swiftly. The average wait statewide is lowest for prenatal genetic patients, which is expected given the time constraints imposed by pregnancy, with an overall average wait of approximately 1 week, although some centers report a 4-week wait time. For general/pediatric genetic clinics, the average wait is slightly over 4 weeks statewide; however, in Northern Virginia, clinics are booked up to 5 months into the future, illustrating a critical manpower shortage in that region (1). Other areas of the state have wait times of up to 12 weeks for pediatric patients. Although most of the large genetic centers in the state have ongoing clinical outreach programs, Virginians living in areas geographically removed from the main genetic centers must travel quite a distance or face significant delays in obtaining genetic care. In addition to clinical sites throughout the metropolitan areas where their centers are located, survey respondents reported participating in outreach clinics in Abingdon, Fredericksburg, Gloucester, Lynchburg, Nassawadox, Newport News, Roanoke, Winchester, and Beckley, West Virginia, most of which are held on a monthly or quarterly basis. The occasional use of telemedicine for clinical genetic services was also reported.

Participants' perceptions of genetic services in Virginia—including quality, accessibility, and manpower issues—were measured by multiple questions throughout the survey. Although 83% of respondents believe the overall quality of clinical genetic services in Virginia is good or excellent, 78% indicated they agree or strongly agree that there are problems with access to services. Most commonly, limited outreach to rural Virginians was cited as a significant accessibility issue, with 61% of respondents agreeing that the

geographic distribution of genetic providers is inadequate to meet current needs. In addition, 78% of respondents indicated that the number of genetic providers currently practicing in the state is not sufficient. Insurance coverage restrictions, low reimbursement, and limited cancer genetic services were also reported by multiple respondents as significant barriers hindering service accessibility. Insurance coverage restrictions for genetic patients were also frequently cited as the reason for limited access to other medical specialists, medications, and recommended laboratory testing, with 82% reporting insurance requirements that result in lab work being performed by laboratories that are not of the highest quality. More than half of respondents cited examples resulting in adverse outcomes for patients, including delayed testing results, increased costs to patients because of test inadequacy, and, even more disturbing, inaccurate test results. The most commonly suggested methods of improving genetic services accessibility in the state were improvement of insurance reimbursement and coverage, increased outreach to underserved areas, more genetic providers, and better education of non-genetic providers and the public.

### Quality Assurance

Based on survey responses, it appears that ongoing performance measures and QA activities vary among genetic centers in Virginia. The most commonly employed QA activities reported include formal and informal case discussion and internal chart review with or without comparison to specific criteria. Periodic review of patient satisfaction data was also reported by a few respondents. The frequency of QA activities also varied significantly, with some daily activities, while others are performed yearly. In a few cases, no organized QA activities were apparent, suggesting the need for education and, perhaps, collaborative efforts in this regard. Participants indicated that several QA activities are particularly helpful, including best practice education, review by outside practitioners, and feedback from referring providers, although these were not reported as current activities at genetic centers in the state. Although not unanimous, most respondents supported standard-of-care guidelines for genetic providers, perhaps because of the difficulty in staying apprised of the burgeoning developments in the field. Despite this, 79% reported being able to stay up to date in genetics. In addition, approximately half of respondents reported sometimes or often enrolling patients in research protocols, suggesting that Virginia's genetic providers are actively involved with continuing education in this rapidly changing field of medicine.

### Available Resources

A variety of resources are critical for the provision of appropriate genetic care, thus their availability throughout the state was assessed in the survey. In particular, teratogen information resources, patient education materials, and cultural and linguistic services were addressed. Most respondents reported weekly use of teratogen references, which are most frequently used in prenatal genetic settings. Multiple sources were reported, with virtually all respondents citing Reprotox ®, an online teratogen database, in addition to other resources. Although not present in Virginia, some states support a centralized teratogen information center or hotline. Although the majority were neutral on this topic, 35% of respondents thought such a service would be a good use of resources for Virginia. Confirming the vital role played by educational resources in providing genetic care, 68% of respondents reported using such resources with their patients often, usually, or always. Multiple formats were noted, including brochures and booklets produced by organizations such as the MOD and genetic support group organizations, as well as scientific articles and Internet resources. Unfortunately, 29% of respondents reported having insufficient patient education resources, largely because educational materials for some genetic conditions or genetic tests are not available or are out-of-date, or because materials are not available in non-English languages or low-literacy formats. The ability to provide culturally and linguistically appropriate genetic services appears to be very limited for many providers in Virginia, with 71% reporting insufficient resources for such care.

Although it varies geographically, 25% of respondents reported that 10% or more of their patients have a first language other than English, most commonly Spanish or Asian languages, although French as well as African and Arabic languages were also reported. The most frequently cited barriers to providing culturally competent care were limited linguistic and cultural diversity among genetic staff in Virginia; limited availability and funding for interpreters, particularly those familiar with medical and genetic terminology and the nondirective counseling essential to genetics; and limited availability of information resources and consent forms in languages other than English. In particular, respondents reported needs for Spanish and Asian— particularly Chinese and Vietnamese—language resources and in-person interpreters.

### Reimbursement Issues

Reimbursement issues remain a critical concern for genetic providers in Virginia and across the country. All survey respondents indicated that Medicaid and Medicaid health maintenance organization (HMO) reimbursement is inadequate, while 79% also cited private insurance reimbursement for clinical genetic services as somewhat or very inadequate. For 67% of respondents, reimbursement is more of a problem for clinical genetics than other services at their institution, suggesting that at least part of the problem is specialty-specific. On average, reimbursement levels were reported to cover only approximately half of the charges billed and about the same percentage of the actual cost of providing services. While some centers reported having other funding sources—such as state support or other grants, institutional support for indigent care, or self-pay patients—many reported no other source of support for clinical services. With an average of almost two hours per week personally spent trying to obtain coverage or reimbursement for genetic services, not including the time spent by administrative staff, 75% of respondents indicated that insurance referral and authorization problems are somewhat or very significant for their genetic center. Respondents most commonly noted genetic counseling, particularly when performed by a genetic counselor instead of a physician or not in conjunction with a procedure, and non-routine genetic tests as non-covered services. Prevention and preconception services and genetic testing for the partners of pregnant females were also cited as problematic reimbursement areas. Although no consistent success was noted, respondents reported using a variety of strategies to address insurance reimbursement problems, most frequently including letters of medical necessity, phone calls to insurers, and meeting with insurance company medical directors. Suggestions for addressing reimbursement issues in Virginia included educating insurers in the state, collaborating with national organizations undertaking advocacy and education, promoting relevant insurance legislation, and increased state support.

### Legal Questions

Genetic discrimination and privacy are issues of concern in Virginia and worldwide, yet survey respondents reported that their patients are largely unconcerned about the possibility of genetic discrimination, suggesting a lack of public awareness or a lack of actual problems in this area. Although most respondents are not aware of patients that have experienced such discrimination, the majority are not satisfied with the level of protection afforded by genetic discrimination and privacy laws currently in place in Virginia. The existing Virginia legislation as well as relevant federal regulations are described in more detail in the previous chapter.

### State Leadership

The level of involvement of state organizations and public health leaders in clinical genetic services varies from state to state, with different administrative arrangements supporting the various levels of the public health pyramid from infrastructure support to direct services. Survey respondents appear to have a

fairly accurate perception of the role of VDH in population-based services such as newborn screening, early hearing detection, and Virginia's birth defects registry. In addition, respondents seem to appreciate the responsibility of VDH to address legislative issues and state educational concerns while also providing monetary support to genetic centers for some patient services. Respondents also expressed concern about the challenges of sustaining support for genetic services and testing for indigent and underserved patients and families in the state, particularly given that genetics has not been a focused priority of the leadership of the health department in the past decade. Respondents encouraged a more active role in all divisions of VDH and other statewide organizations in assessing and addressing the needs for genetic services and education across the lifespan, and in facilitating the development of a comprehensive infrastructure for genetic services that provides universal access for patients. Respondents also support an increased role for VDH in efforts dealing with reimbursement and genetic discrimination issues, as well as statewide education initiatives and improvement of coordination and collaboration among stakeholders.

### Educational Needs

Gaps in the genetic knowledge and awareness of non-genetic healthcare providers have been noted throughout the country. This perception is also true in Virginia, where 67% of survey respondents are dissatisfied or very dissatisfied with the level of Virginia's non-genetic healthcare providers' understanding of genetics and clinical genetic services, while a similar number indicated that the general medical community only rarely or sometimes has adequate training to provide appropriate care for patients with birth defects and genetic conditions. While respondents indicated that most referrals for genetic services are made appropriately, many are concerned about significant under-referrals of patients that should receive genetic specialty care. In addition, most genetic referrals are from health departments, PCPs, and pediatric and obstetric specialists, with few from other adult specialists, suggesting that some medical specialties may be particularly unaware of the genetic needs of their patients or the benefits of genetic services. To improve the appropriateness of genetic referrals, survey respondents frequently suggested increased education and marketing to PCPs and specialists, perhaps including a genetic hotline; the distribution of genetic referral guidelines; and public education. Respondents noted that several medical groups are particularly in need of education about genetics, including (1) PCPs, nurse practitioners and midwives; (2) adult specialists; (3) health department providers; (4) early intervention service providers; and (5) child abuse teams. Also cited were several non-medical professional groups, including insurers, legislators, and reporters. Despite this acknowledged need, 33% of respondents are involved with the education of Virginia's non-genetic providers once per year or less, with an additional 50% participating in such educational endeavors only 2-5 times per year, further suggesting that high clinical workloads limit other endeavors for Virginia's genetic providers. For the most part, the educational activities with which respondents are involved include lectures and inservices targeting medical residents, medical students, nursing students, and staff in academic medical centers. While some outreach education of health department and community providers was also reported, these results indicate a large gap in genetic education efforts targeting providers in many of Virginia's communities.

The survey results also suggest significant gaps in the public's knowledge of genetics. While 63% of respondents reported being dissatisfied or very dissatisfied with Virginians' understanding of genetics, 76% were dissatisfied with their awareness of clinical genetic services. Unfortunately, respondents' participation with public genetic education is quite limited, with 67% reporting being involved with such efforts only once per year or less. These results provide significant insights into barriers to the provision of appropriate genetic care and illuminate targets for future development.

# 6. PAYING FOR GENETIC SERVICES: A REVIEW OF VIRGINIA HEALTH INSURANCE ISSUES

## The Origins of Health Care Plans

Problems with the financing of genetic services present significant barriers to the provision of appropriate genetic healthcare in the U.S. Although some of these issues are fairly unique to genetics, others are broad-based and have their roots in the evolution of the U.S. health care system over the past few decades. Insurance and issues of the uninsured have developed over the past century as the insurance industry has become a complex system that heavily emphasizes managed care with its goal of maintaining a healthy population while containing costs.

In the late 1920s, a few doctors offered their services to the workers and families of various organizations for a predetermined fee. Henry Kaiser first opened up his prepaid medical plans to the public after World War II. By the mid 1950s, Kaiser Permanente, managed by the Kaiser family and company officers, had over a half million people enrolled in its health plan, with access to Kaiser's own clinics and hospitals. Other types of prepaid insurance plans had also evolved by the end of the 1940s. Management and decisions were made by a board of representatives or by the enrolled members through elected trustees. The earliest example of an independent practice association (IPA) prepaid health plan, controlled by a medical foundation, dates back to the mid 1950s. Initially, the American Medical Association (AMA) did not support any of these forms of "organized" medicine and they remained a small presence in the health care world until the early 1970s (1).

Rising costs and concerns about new therapies and appropriate care led to an emphasis on a new type of health plan, the Health Maintenance Organization (HMO), which was first supported by the Nixon Administration in 1971. The HMO Act of 1973 authorized $375 million in federal funds to help develop HMOs, preempted state laws that banned prepaid groups, and required companies with at least 25 employees to offer a federally qualified HMO health insurance plan to its employees. The ultimate goal was to have 90% of the U.S. population enrolled in an HMO by 1980. As a result of this support, over 600 HMOs were in existence by 1996 (1).

Though the initial planners' goals regarding population enrollment have not been met, managed care health plans have become the norm. Following much consolidation and adaptation over the past 30 years, these plans continue to vary in their capacities to control costs, provide comprehensive services, and enhance quality of care. Currently, there are three main types of managed care health plans in practice. HMOs and Point of Service Plans (POS) are the most prevalent, with the insurance company as the active financial manager for both types. They restrict patients' choices and are actively involved in negotiating contracts for payment of doctors and hospitals. A designated PCP is the "gatekeeper" for each patient's care and treatment. Preferred Provider Organizations (PPO) are the third type of managed care health plan. PPOs tend to be the least restrictive, with contracts negotiated directly between employees and insurers. Though there is broader access to care, there are generally more "out-of-pocket" expenses incurred by the insured individual. Obviously, managed care today has evolved into a large, competitive, complex business. It provides a system of health care delivery that tries to control the costs and quality of health by managing access to medical care.

Unfortunately, many people across the country are still not covered by any health care plan, and the "underinsured" represent a growing concern as well. The latter group includes insured individuals with chronic genetic conditions requiring lifelong care in which insurance expenditure limits fall far short of the cost of care. In other cases, insurance coverage facilitates the diagnosis of a genetic condition, but

does not cover the costs of treatment. Total enrollment levels in private and government health plans compared to the levels of uninsured individuals in Virginia and nationwide are shown in Table 6-1, while the number of children ≤ 18 years old in each category are detailed in Table 6-2 (2).

**Table 6-1. Types of Insurance Coverage**

| Insurance Provider | Virginia Coverage Levels 2003-2004 | | National Coverage Levels 2004 | |
|---|---|---|---|---|
| Employer policy | 4,342,350 | (59%) | 155,778,670 | (54%) |
| Individual policy | 339,770 | (5%) | 13,968,130 | (5%) |
| Medicaid | 537,950 | (7%) | 37,242,750 | (13%) |
| Medicare | 868,870 | (12%) | 34,379,930 | (12%) |
| Other Public | 213,940 | (3%) | 3,096,400 | (1%) |
| Uninsured | 1,011,420 | (14%) | 45,820,480 | (16%) |
| Total | 7,314,310 | (100%) | 290,286,350 | (100%) |

**Table 6-2. Insurance Coverage for Children**

| Insurance Provider | Virginia Coverage Levels 2003-2004 | | National Coverage Levels 2004 | |
|---|---|---|---|---|
| Employer policy | 1,256,610 | (65%) | 43,803,740 | (56%) |
| Individual policy | 77,620 | (4%) | 3,396,530 | (4%) |
| Medicaid | 335,110 | (17%) | 20,514,050 | (26%) |
| Other Public | 81,240 | (4%) | 1,045,500 | (1%) |
| Uninsured | 169,360 | (9%) | 9,037,120 | (12%) |
| Total | 1,919,940 | (100%) | 77,796,940 | (100%) |

## Determinants of Health Care Coverage

Cost benefit analysis attempts to measure the costs and benefits of a service or product to patients, families, and society and then converts that to a common unit, such as the dollar. Cost effect analysis uses non-monetary measurements and often compares alternatives that result in a comparable health benefit such as years of life (3). Cost effect analysis is generally preferred and may be particularly applicable to genetics. This type of analysis involves assessing and evaluating issues such as efficacy of treatment, effectiveness of screening, prevalence of a disease, and the likelihood of a mutation (4). While these economic parameters sound reasonable, they are very difficult to measure. Insurance providers are challenged to determine medical necessity and ration services. By definition, managed care plans are expected to provide effective services and contain costs, while for-profit plans must also generate a profit. Though resources are limited and the ultimate goal is to improve and maintain the health of the population, there are many individuals with disabilities and special needs who require regular and frequent access to health services throughout their lifetime. This results in a need to individualize the determination of medical necessity and assure that continuing medical coverage is made available to this population. Health insurance providers, consumers, and the government are variably addressing the challenges of providing services to these populations. The benefits of various insurance packages for children with special health care needs in Virginia are currently being examined through a federal grant

(5). Combined with the ongoing development of new genetics tests and therapies, individuals with genetic conditions are frequently challenged by insurance coverage decisions, regulations, and policies that do not change readily and are often not designed to address specific needs.

In Virginia, only supplies, equipment, and appliances that are considered medically necessary must be covered by insurance plans. Medical necessity statements from commercial insurers vary, but generally include services that meet the basic health needs of a person, are rendered in the most cost-efficient manner, are consistent with the diagnosis and predetermined guidelines of treatment for a condition, are required, and are safe and effective (6).

### Coverage of Genetic Services

Medical necessity guidelines are applied to genetic coverage in various ways by different insurers. The current needs assessment process included an evaluation of the coverage of genetic services by the ten largest providers of health insurance in Virginia, as determined by the Bureau of Insurance of the Virginia State Corporation Commission. Results are summarized in Table 6-3. Actual levels of reimbursement by these insurers vary. During the assessment process, it was found that most insurance companies' customer service providers and provider relations representatives did not know all of the details of their own companies' genetic policies, nor was this information always available in member handbooks. Most of the information was able to be obtained through evaluation of the policies on the insurance companies' Web sites, however. This highlights the essential need for providers and consumers to be familiar with appropriate terminology in order to fully access available coverage for genetic services. As noted in table 6, all of the policies provide coverage for consults with either a clinical geneticist or a genetic counselor under certain circumstances, in addition to coverage for amniocentesis testing. Most also allow for other genetic diagnostic or screening tests for prenatal patients, illustrating a fairly universal understanding of the importance of good prenatal care, which sometimes requires genetic services.

Of particular note, Anthem's genetic medical coverage guidelines include a detailed description of the utility of genetic testing, stating

> "genetic testing involves several different types of testing to determine if a person has a genetic mutation known to cause certain diseases. The ability to identify genes associated with certain diseases contributes to many facets of medical care. Genetic testing can be used to identify patients who are at significant risk of developing life-threatening illnesses. Identification of high-risk patients may allow them to make informed decisions about potential pre-emptive therapies and other medical options. Genetic testing also plays an important role in identifying diseases difficult to diagnose. This role can lead to the avoidance of unnecessary medical tests and treatments. Further, for many conditions proper diagnosis may allow appropriate treatment to begin earlier. Finally, genetic testing of couples planning to have children may alert the risk of giving birth to a child with a genetic defect known to cause certain diseases."

The guidelines also discuss the fact that genetic testing may impact other family members, and stress the importance of discussing all details of the implications of testing with a trained genetic professional such as a genetic counselor. In addition, Aetna's clinical policies provide very specific and detailed guidelines regarding genetic counseling, all types of genetic testing, and prenatal care.

## Table 6-3. Coverage of Genetic Services in Virginia

| *Insurance Company* | *Genetic counseling* | *Genetic consult with MD specialist* | *Diagnostic genetic testing* | *Screening genetic testing* | *Amniocentesis* | *Method of genetic testing coverage determination* | *Comments* |
|---|---|---|---|---|---|---|---|
| 1. Anthem* | Yes | Yes | Yes | Yes | Yes | If medical necessity after genetic counseling and clinical exam | http://www.anthem.com<br>May need authorization/PCP approval |
| 2. Optima | No | Yes, if medical necessity | No | No | Yes, if medical necessity | Medical care management review and PCP authorization | |
| 3. Health-keepers* | Yes | Yes | Yes | Yes | Yes | If medical necessity after genetic counseling and clinical exam and authorization/PCP approval | http://www.anthem.com |
| 4. Optimum Choice | Yes, prenatal | Yes | Details not available | Yes, prenatal | Yes, if medical necessity | Services provided based on group plan and/or what benefits/ coverage an employee selects | http://www.mamsi.com<br>Over 1000 plans with varying policies; see guideline for routine prenatal care |
| 5. Carefirst | Yes | Yes | Yes, if medical necessity | Yes, if medical necessity | Yes | Depends on the individual plan/policy and PCP authorization | http://www.carefirst.com<br>Many different plans; see medical policy reference manual |
| 6. Southern Health | Yes | Yes | Yes | Yes | Yes, age ≥35 or with high risk of Down syndrome | Coverage for birth defects; PCP evaluates family history; preauthorization for genetic counseling and testing required | http://www.southernhealth.com<br>Authorization necessary if specialist is non-participating; see clinical preventive services |
| 7. Kaiser | Details not available | Details not available | Generally ok | Generally ok | Details not available | Testing covered if medically necessary and covered under selected plan and selected options within the plan | |
| 8. Aetna | Yes | Yes | Yes, if required conditions met | Yes, if required conditions met | Yes, if medical necessity | Dependent on plan/policy of individual and authorization of PCP | http://www.aetna.com<br>See clinical policy bulletins: genetic counseling, genetic testing, and prenatal diagnosis. |
| 9. Priority* | Yes | Yes | Yes | Yes | Yes | If medical necessity after genetic counseling and clinical exam | http://www.anthem.com<br>May need authorization/PCP approval |
| 10. VA Premier | No | Yes, for pregnant patient | Yes, for diagnosis of a fetal condition | No | Yes, if previous procedures determined necessity | If patient is pregnant and there is concern about problems with the fetus | |

* All Anthem guidelines apply

The field of genetics is rapidly growing and changing, while simultaneously competing with other health specialties for the valuable insurance dollar. As more genes are discovered and linked to disease, the importance of providing genetic services and testing is becoming increasingly evident. When consideringwhether or not to provide coverage for genetic services, health insurers must consider several factors: (1) who are the patients and how will they be affected; (2) is there consumer, patient, or provider demand; (3) what are the costs of the service and the costs of providing the service; (4) is there a great potential for cost savings in the future if coverage is provided; (5) is this mandated by the state; and (6) will providing coverage result in being more competitive (7). These questions should also be considered as plans for genetic programs and initiatives to address reimbursement gaps are developed.

Clinical genetics, a relatively new and small medical specialty, is a multidisciplinary field that includes MD's; PhD's with clinical, biochemical, molecular, and cytogenetic backgrounds; and genetic counselors as service providers. These providers often work as a team in order to provide all of the necessary services to patients. This team approach is somewhat unique and is often not fully recognized by insurance companies. As a result, insurance reimbursement for services often does not reflect the time and complexity of history-taking, physical assessment, literature review, appropriate diagnostic testing, and follow up.

Knowledge of genetic disease is important to both the PCP and the medical manager of an insurance company. Timely and appropriate genetic referrals are essential. Despite the fact that genetic testing can be quite expensive, dollars can actually be saved with appropriate testing. With an accurate, specific diagnosis, correctly targeted care and management can begin, which can ultimately lead to substantial financial savings. In addition, uncertainty and stress for patients and their families can be reduced, with potentially far-reaching impacts within families. Unfortunately, health insurers have historically been more willing to pay for the costs of illness than for the cost of health, although increasing emphasis has been placed on preventive care in recent years (8). Insurance companies, particularly managed care plans, must carefully assess the overall benefits of genetic care, including its role in preventing disease and minimizing morbidity, however, knowledge of genetics is often not high in the insurance industry. One exception is the Kaiser HMO in California, which hires a genetic counselor for every 80,000 enrollees and a clinical geneticist for every 350,000 to 400,000 enrollees. Databases provide coordinated information and some services can be effectively delivered over the phone as billing is not done for individual visits (5). Such programs provide models for future service planning.

Funding for genetic services comes from a variety of sources other than insurance providers, a necessity since insurance reimbursement, particularly Medicaid, generally falls far short of paying for the actual cost of genetic care. These coverage shortfalls have become magnified as insurers have increasingly contracted with PCPs and hospitals for inpatient and outpatient care, and with large commercial laboratories for medical testing. The contract restrictions often make genetic diagnosis difficult for patients and genetic providers because many genetic tests necessary for definitive diagnosis are uncommon, may not be offered by contracted laboratories, and frequently, are only available through out-of-state laboratories. Other funding sources for genetic services include grants, such as the MCHB Title V Block Grant. This grant was authorized by the 1935 Social Security Act to provide health care to all mothers and children. The funds are given to states, which distribute them to various organizations in order to meet determined goals. Funding for some genetics services may also be available through other sources, such as research grants and grant funds from specific disease organizations (9).

Obviously, there is a need for providers and consumers of genetic services to work with state and federal governments and the insurance industry to assure the provision of optimal genetic healthcare. The limitations of procedure and diagnostic coding, poor coverage of genetic testing, the disallowance of

billable services by genetic counselors, and insurance discrimination are all problems that need to be addressed. In addition, with the increasing lifespans of individuals with birth defects and genetic conditions, issues regarding their transition into adulthood will continue to emerge. As the number of genetic tests and the need for genetic expertise in many aspects of medicine grow, these insurance gaps will become even more critical. All of these concerns highlight the need for a significant overhaul of genetic healthcare coverage.

# 7. NON-GENETIC HEALTHCARE PROVIDERS: CURRENT PRACTICE AND FUTURE ROLES IN GENETICS

A growing understanding of medical genetics is expected to have an increasingly significant impact on the diagnosis and management of disease. In fact, the medical profession is thought by many to be in the early stages of a revolutionary change toward a genetic perspective of disease, with a shift in emphasis from the passive treatment of symptoms to the active prevention of disease through the combined use of predictive genetic testing and preventive therapies based on an individual's genetic makeup and improved understanding of many gene–environment interactions. Given these expectations of expanded insight, the ability of the health workforce to apply the new knowledge to patient care has come under intense scrutiny. For many years, policymakers have commented that the genetic provider workforce is a fraction of the size necessary to cope with the expected patient load, although efforts to expand that workforce have been limited. One widely advocated solution is to have PCPs and non-genetic specialists provide a significant portion of genetic services. While virtually everyone agrees that improved genetic knowledge of all health professionals is necessary, some question whether such a system is feasible, particularly given existing constraints within primary care, as well as the fact that, despite years of effort by genetic providers, reimbursement for genetic services generally remains insufficient. Of particular concern is the possibility that PCPs without sufficient training will provide inaccurate risk assessment, will choose inappropriate tests, and will misinterpret DNA test results (1, 2). At the least, patient demand for genetic information will result in PCPs and non-genetic specialists increasingly being called upon to serve as gatekeepers of genetic services, with expanded risk assessment capabilities necessary for appropriate referral (3). Unfortunately, very little information is available regarding the genetic services provided by PCPs and other non-genetic providers in Virginia, or about their attitudes toward future changes in service models. While findings regarding providers in other states and abroad may not entirely reflect conditions in Virginia, they provide general insights that may be instructive for future planning, and that general data is summarized here. The results of the Virginia PCP survey conducted as part of this needs assessment process are summarized in the next chapter.

## Traditional Roles and Interactions

Currently, among non-genetic providers, pediatricians and obstetricians generally are the most aware of genetic services, largely due to their increased level of exposure to the many genetic conditions that present in childhood or are subject to prenatal testing. Adult providers tend to be less aware of these services, perhaps because many of the more severe genetic disorders are often not compatible with survival to adulthood. The roles of various providers caring for patients with single gene or chromosomal disorders are fairly well established, with the PCP, usually a pediatrician, raising the initial concern about a genetic condition. Pediatricians in particular are well-suited to identify and initially evaluate genetic conditions and developmental delays in young children because they are generally familiar with the perinatal and family history and provide longitudinal care during which problems with development can be recognized (4). A pediatric genetic provider can then be consulted to facilitate diagnosis and genetic counseling. Traditionally, the genetic provider and PCP provide ongoing management of the patient collaboratively, with additional specialists involved in the care of patients that have conditions or symptoms related to their area of expertise (5). Some pediatricians are reluctant to provide long-term management of chronic conditions, citing issues of time and reimbursement as well as the fact that their training has traditionally focused on preventive health maintenance ("well child care") and acute pediatric illnesses. However, pediatricians often serve a critical role in care coordination (4). The AAP actively promotes the preparation and publication of practice guidelines for children with common genetic disorders to facilitate appropriate ongoing management through the medical home (6). Although not exhaustive, Table 7-1 indicates well-established referral

indications for pediatric genetic services (7).

**Table 7-1. Common Referral Indications for Pediatric Genetic Services**

| |
|---|
| Two or more major congenital malformations or one major plus several minor malformations |
| General dysmorphic appearance |
| Mental retardation, especially in conjunction with metabolic disturbance, congenital malformation, or a positive family history |
| Neonatal lethargy or coma, or other metabolic disturbance without explanation |
| Loss of milestones, organomegaly, or both after initially normal development |
| Abnormal sexual development, primary amenorrhea, or aspermia |
| Abnormal newborn screening result |
| Unexplained loss of hearing or sight |
| Unexplained short stature or abnormal pattern of growth |
| Neurodegeneration |
| Unexplained abnormalities of the skin, hair, or bones |
| Blood disorders such as sickle cell disease or coagulopathy |
| Immune deficiency |

Obstetrician-gynecologists frequently interact with genetic providers when they discover elements in a pregnant patient's history—maternal age, abnormal screening tests, family history, potential teratogenic exposures, or maternal disease—that raise concerns about the possibility of a birth defect or hereditary condition in the pregnancy. Such patients are generally seen by a genetic provider for risk assessment, genetic counseling, and diagnostic testing when available, while ongoing pregnancy care is provided by the obstetrician (5). While couples with a family history of various hereditary disorders have sought genetic risk assessment for many years, the development of prenatal diagnostic techniques, such as amniocentesis as well as the growth of molecular genetic testing, have vastly increased the clinical applicability of this area of genetics. Offering prenatal testing for genetic conditions due to maternal age or other risk factors became the standard of care in the 1970s following several lawsuits by patients at risk that had not been offered such testing (8). The Collaborative Ambulatory Research Network (CARN) is an effort of the American College of Obstetrics and Gynecology (ACOG) to identify practice patterns and educational needs among its fellows working in an ambulatory care setting. CARN assessments during the late 1990s indicated that the use of family history for risk assessment and DNA-based testing is more common in obstetric than gynecologic practice. Eighty-six per cent reported using genetic history forms for prenatal patients and a similar number reported having access to genetic counselors; however, a significant proportion indicated that they provided genetic counseling and prenatal diagnosis themselves rather than referring to a genetic provider (9). Family practitioners caring for pregnant patients may interact with genetic providers under similar circumstances; however, in practice this tends to occur less often than with obstetricians (5).

Other medical specialists interact with genetic providers to varying degrees, often depending on their interest and knowledge, yet articles regarding genetic issues regularly appear in the medical literature of virtually all specialties. Unfortunately, some specialists do not recognize the relevance of genetics to their daily clinical practice, an educational barrier that may be difficult to overcome (10). For patients with common conditions that can be of genetic origin—including cancer, hypercholesterolemia, diabetes, hypertension, and others—the roles of various providers are not well established. Differentiation of the hereditary forms of these disorders from the more common cases where inherited factors have less influence can be difficult because the presentations are usually quite similar. Family history and early age of onset are generally key indicators of the hereditary forms. The practice patterns of the PCPs and

specialists caring for these patients vary somewhat, with some incorporating tools that facilitate the recognition and subsequent referral of the high-risk patients for further evaluation, while others do not. In some cases, the PCPs and non-genetic specialists involved provide genetic counseling and testing themselves without input from genetic providers, with mixed results as discussed in the next section. For the high-risk patients and families that remain unrecognized, the results can be devastating because without recognition they are not offered surveillance or preventive treatment (5).

The scope of genetic practice has increased substantially over the past decade with the discovery of several genes involved in cancer, particularly BRCA1 and BRCA2. These developments provide a glimpse of potential future models of care, as well as pitfalls to avoid. As these cancer genes were discovered and their role in cancer causation elucidated, programs of risk assessment using pedigree analysis and genetic testing were developed. For the most part, these programs have been developed by interested oncologists and genetic counselors, and are housed within oncology or internal medicine groups, with peripheral involvement of clinical geneticists, although in some cases, geneticists play a major role (1, 11, 12). The American College of Medical Genetics (ACMG) responded to these changes by developing guidelines for the counseling and genetic testing of patients with a family history of breast or ovarian cancer or a similar indication. These guidelines support the role of the PCP in identifying high-risk patients by taking a pedigree, with further involvement in the complex and time-consuming genetic counseling left to the discretion of the PCP (13). The American Society of Clinical Oncology (ASCO) and the U.S. Preventative Services Task Force have also issued policy statements regarding genetic testing for cancer susceptibility (14, 15). Once again, however, the success of these programs depends on appropriate recognition and referral of at-risk patients by the PCP or non-genetic specialist. Cardiology is another area in which an expanded genetic role is beginning to emerge in clinical practice, with practitioners increasingly relying on genetic studies to elucidate disease mechanisms, confirm clinical diagnoses, and guide management. The interaction of genetic providers with cardiologists continues to vary significantly throughout the country, however (16).

### Primary Care Providers: Knowledge, Attitudes, and Gaps in Care

The impetus for increasing the role of PCPs in genetic services is supported by several factors. The number of PCPs relative to genetic providers is a key issue. In addition, PCPs generally establish long-term relationships with patients and often use a family-centered approach, both of which are considered beneficial elements of genetic care. Because of this long-term relationship, patients usually direct genetic questions to their PCPs (17). Family medicine education traditionally includes the use of the “genogram,” a type of pedigree in which psychosocial issues and family dynamics are taken into account, information that is also useful in genetic counseling. Follow-up visits to discuss test results are already a common practice and one that could prove useful for a comprehensive discussion of issues relevant to genetic susceptibility testing. The family-centered long-term approach of most PCPs also facilitates addressing genetic issues as they become relevant over a lifetime. Unfortunately, malpractice litigation and the demand created by direct marketing of genetic testing to consumers may influence genetic practice among PCPs as much or more than any planned initiatives (1). Particularly as the number of predictive genetic tests and the availability of preventive treatments increase, the use of genetic information for preventive medicine will rise, an application of genetics well suited to the traditional role of PCPs (3).

Impediments to an increase in genetic service provision by PCPs and non-genetic specialists are also significant. The most obvious is a lack of knowledge about genetics and genetic services (18,19). A recent survey of U.S. medical schools by the Association of Professors of Human or Medical Genetics indicated that the average medical student received a total of 29 hours of didactic genetic coursework, while past students received much less genetic instruction (20). Even with a drastic overhaul of genetic training

within medical education, the proportion of new graduates in the workforce is very small and thus would have little effect in the short-term, indicating a need for continuing education programs in genetics for providers already in practice. Several studies have documented an insufficient familiarity with genetics among PCPs. A study of PCPs in the Pacific Northwest indicated that only 25% of internists and 31% of family practitioners had referred a patient for genetic consultation within the past year. More disturbing was the finding that 1 in 6 internists were unaware of genetic services in the region and saw no need for such information. Over half of internists and obstetricians did not recognize that multiple relatives with breast cancer on the paternal side of the family would increase a patient's risk of disease, yet most of these practitioners reported that they would provide risk counseling themselves rather than refer to a genetic specialist (21). The CARN survey of obstetrician-gynecologists indicated that while almost all respondents asked their prenatal patients about teratogenic exposures—such as smoking, alcohol, and drug use—many did not follow through with referrals or other behavior modification supports (9). Such results indicate that knowledge alone is not necessarily sufficient to effect improvements in practice. In another survey of obstetrician-gynecologists, 21% of respondents noted that they were the only provider of genetic services for their patients, yet 65% did not consider themselves well informed about genetics (22). The situation was similar in a survey of allied health professionals, in which 70% reported discussing genetic issues with patients, although 80% of these professionals had no genetics training (23). In some cases, PCPs perceive an adequate knowledge of genetics, yet actually perform quite poorly on assessments (24). Limited awareness of genetic services seems to be a critical issue for many groups, as exemplified by a national survey of oncology nurses, which indicated that only 35% were aware of referral resources although almost half had received recent patient inquiries regarding cancer genetics (25). The rapid expansion in genetic knowledge and the availability of genetic tests also serves as a barrier for many non-genetic providers, as exemplified by an Arkansas survey, which revealed that only 21% of PCPs, gastroenterologists and hematologists were aware of the option of genetic testing for hemochromatosis one year after it became available as a routine clinical test (26). Advocates for an increased involvement of PCPs and non-genetic specialists in the provision of genetic services promote educational efforts to remedy this lack of genetic knowledge; however, some of these providers must still be convinced of the need to become better educated, as they are reluctant to provide such services at all (27).

Assessments of the attitudes of PCPs and non-genetic specialists toward genetics show variable results. In many cases, it appears that genetics is perceived as an important area of medicine and one in which primary care should play a role. A lack of knowledge and significant educational needs in genetics are widely agreed upon; although, some have suggested that PCPs will be unlikely to respond to educational efforts until they perceive genetic issues as relevant to their current practice, as opposed to a future practice need (18, 28, 29). Some PCPs and non-genetic specialists believe that the clinical utility and validity of some genetic tests have not been well established, contributing to their reluctance to change their current practice (29, 30). A nationwide survey of physicians of various specialties indicated that many believe that genetic tests for cancer susceptibility yield too many inaccurate and ambiguous results, while the majority indicated that clear management guidelines for patients with positive test results are not available. Such results highlight a need for education about the intricacies of genetic testing and clarification of management guidelines. Interestingly, while 84% of oncologists felt qualified to recommend genetic testing to their patients, only 48% felt qualified to provide cancer genetic counseling (31). In addition, several surveys of PCPs have revealed a growing discontent with the increasingly specialized roles being required of them, contributing to concerns about workload (32). A variety of service improvement mechanisms have been proposed by PCPs, including further development of referral guidelines, computerized risk assessment tools, and community genetics clinics supported by the larger genetic centers (33, 34). Overall, PCPs seem to favor a future role that largely involves functions with which they are already familiar, including family history taking and risk assessment to facilitate

appropriate referral, as well as supportive counseling, although even within these role limitations, practice improvements are necessary (18, 32).

Eliciting and interpreting family medical histories is an important tool in all areas of medicine, particularly regarding genetic conditions. Risk assessment in primary care has long relied on these tools for appropriate patient triage and specialty referral, while the ongoing incorporation of genetic advances into clinical care will undoubtedly make these functions even more vital in the future. Unfortunately, many studies indicate that the elicitation and interpretation of family histories is often not complete or appropriate in primary care and other non-genetic specialties. A survey of obstetrician-gynecologists revealed that 76% do not solicit family history as part of a typical gynecology visit, which is frequently the only regular health care obtained by women in this country, particularly those of reproductive age (22). Preconception assessment of genetic risks is considered preferable to prenatal assessment and, frequently, allows for interventions that lower risk, yet PCPs often do not elicit family histories preconceptually, thus undermining this potential for preventive care (35). A Maryland study involving a review of prenatal records revealed that in 36% of patients, obstetric care providers, although they had obtained a family history, failed to note a significant genetic risk that was subsequently elicited during genetic consultation for other indications (36). A large British study of families with a known risk factor for one of several avoidable genetic conditions revealed that a large percentage had not been counseled by their PCP or offered a referral to a genetic provider, resulting in disease occurrence (37). A study of Ohio family practitioners indicated that family history was discussed in only 51% of new patient visits and 22% of established patient visits (38). In another study, the percentage of appropriate assessments of genetic risk based on several hypothetical family histories by general practitioners in England ranged from 21% to 63%, while the percentage of appropriate referral decisions ranged from 40% to 80% depending on the family history (39). Other studies have noted that a significant proportion of genetic referrals are of low-risk patients (40). These findings suggest that the use of family history to recognize genetic risks is being underutilized by PCPs currently and that risk assessment abilities are limited in this group, supporting the development of protocols for improvement of family history assessment by PCPs. Some have proposed that practice nurses may provide some basic genetic assessment in primary care; however, significant educational needs have been identified among this group as well. A survey of nurses in Great Britain indicated that while the vast majority routinely collects family history information, many are not confident about collecting relevant details regarding familial cancer, and are even less certain about how to assess the history information (41). A survey of general practitioners revealed similar results, with most believing they should play some gatekeeping role involving family history assessment, yet lacking confidence in their ability to perform this function (42).

Inappropriate interpretation of genetic test results by non-genetic providers is also a significant concern. In an Ohio study, family practitioners, internists, and gynecologists were asked to interpret a BRCA1 / BRCA2 genetic testing report for a hypothetical patient. Although all participants correctly identified the patient's mutation status, the patient's resulting cancer risk was inappropriately interpreted by many respondents, and one third did not have sufficient knowledge of Mendelian inheritance to correctly calculate the chance that relatives would inherit the altered gene (43). A study of APC testing for familial colon cancer indicated that in almost 1/3 of cases, the physician incorrectly interpreted test results (44). Genetic test results are likely to only become more complex and; therefore, more prone to misinterpretation by providers unfamiliar with them, as multiple genes, each with multiple possible mutations with different effects and variable interactions, are found for common disorders that may also be affected by environmental exposures (2). Such misinterpretations of disease risk and implications for relatives could have disastrous consequences, including inappropriate surveillance and missed opportunities for intervention in at-risk patients (43).

A central tenet of genetic counseling has been the non-directive approach, particularly regarding reproductive issues. Much of medical practice outside of genetics is fairly directive, however, raising concerns about the possibility that PCPs and non-genetic providers will provide directive genetic counseling. A survey of U.S. PCPs and genetic providers revealed that PCPs were much more likely to be directive when presented with a scenario involving a couple's consideration of prenatal diagnosis after being identified as carriers of cystic fibrosis (45). In Great Britain, a survey of obstetricians and genetic providers revealed that the obstetricians advocated a more directive approach to counseling for patients with an abnormal result on amniocentesis (46).

Other major concerns regarding the adoption of the primary care model of genetic service provision revolve around ethical, legal, and social issues. A study reviewing the circumstances surrounding patient samples sent for APC testing for familial colon cancer revealed that less than 20% of patients had given written informed consent or received formal genetic counseling prior to testing (44). Only 2.3% of CARN study respondents indicated that they consider the possibility of insurance company discrimination when ordering genetic tests, while 14% reported obtaining informed consent for DNA tests (9). A survey of family practitioners revealed that they were more willing than geneticists were to disclose information about genetic status to third parties without the patient's consent (47). Despite these findings regarding practice, a nationwide survey of physicians indicated that most are concerned about genetic discrimination and the confidentiality of genetic test results (31). Another consideration is the focus of most PCPs on the individual patient, to the exclusion of other family members. Outside of genetics and perhaps family practice, most healthcare providers do not routinely include a patient's family members when considering the implications of a diagnosis. This omission has already led to litigation in some states, including Florida, where the court found a surgeon liable for not informing his patient of the hereditary nature of her cancer, which subsequently affected her daughter as well, in *Pate v Threlkel*. Standards of reasonable care, informed consent, duty to inform, as well as confidentiality are likely to be changed by the incorporation of genetics into all fields of medicine (2).

The growing push within primary care to see more patients in less time is also not conducive to educating patients about complicated genetic issues, providing non-directive counseling, or facilitating truly informed consent. Particularly in primary care, the time involved in a typical physician-patient encounter is shrinking (48). PCPs in Alabama cited a lack of time as a major reason for not discussing genetic issues (49). Although patient demand for genetic information is certain to increase and some PCPs will want to keep abreast of current developments, the financial incentives for PCPs to provide genetic services are limited. The time commitment involved in the provision of genetic services, particularly genetic counseling, is significant and is poorly reimbursed by insurers. Without adequate time, appropriate attention to the psychosocial, ethical, and legal issues associated with genetic testing is also unlikely (2). Along with the pressure on PCPs to increase patient volumes and thereby reduce costs, they are being asked to expand their roles in many other ways, increasing their reluctance to take on the responsibility of genetic issues as well (27).

### Models for Integrating and Improving Care

Several projects aimed at improving the genetic care provided by PCPs and non-genetic specialists have been tried, with varying success. Many of these can serve as informative models of genetic service integration into primary care. A large number of studies have involved educating key members of the primary care team to provide specific types of genetic services. The Michigan Department of Health coordinates a statewide program of genetic services in which several genetic counselors serve as outreach coordinators and educators, with nurses in many pediatric care settings trained to facilitate comprehensive genetic care and family-centered assessment on the local level (50). A program in Pennsylvania involves

training family-planning personnel throughout the state to provide preconceptual genetic risk screening and education, as well as referral for genetic services when appropriate, with significant success in providing assessment for traditionally underserved populations (51). Several studies have shown that genetic screening for common Mendelian diseases can be successfully integrated into primary care programs (52). An educational effort in Alabama revealed that PCPs can provide improved screening for disorders, such as hemochromatosis following targeted educational initiatives (53). A similar program in New York demonstrated that PCPs could provide appropriate hemoglobinopathy counseling following training by genetic providers, relieving demand on the genetic center (54). A number of studies have evaluated cystic fibrosis carrier screening in primary care, with mixed results. Uptake rates have generally been similar following counseling by PCPs and genetic specialists, although patient misunderstanding of results appears to be a common problem (18, 55).

A variety of initiatives to improve family history assessment in primary care have also been evaluated, with results indicating that appropriate assessment is feasible following relevant training; however, most of these have involved small groups of PCPs interested in genetics and thus may not reflect their utility following broad application (56). In November 2004, the U.S. Surgeon General launched the *Family History Initiative* and introduced an online tool, *My Family Health Portrait,* to encourage all Americans to learn about their families' health histories as a way of promoting personal health and preventing disease (57). Such public initiatives may facilitate an improvement in the use of family history information by PCPs by creating a better-informed public. A pilot project in Wales served to improve the appropriateness of cancer genetics referrals by providing specific referral guidelines for PCPs and a detailed family history questionnaire completed and mailed in by patients prior to being seen by genetic providers (58). In the United Kingdom, several studies are assessing the use of community genetic counselors as outreach workers acting as liaisons between the genetics clinics and general practitioners. The community genetic counselors filter referrals to the genetic centers and serve as educators. Unfortunately, widespread adoption of such a model is unlikely given the limited number of genetic counselors available (3, 59).

The use of computer programs as tools for the provision of a variety of genetic services in the primary care setting have been developed. In some cases, computer tutorials facilitate patient education about genetic testing for cancer susceptibility (60). Other studies have evaluated the use of computer programs for the assessment of cancer genetic risk in primary care, with results revealing some concern about the dynamics of patient consultations involving computer support, although the software was thought to be user-friendly (18, 61). Educational materials are also critical for reinforcing information provided in genetic counseling, and may be even more critical for genetic services provided within primary care. An evaluation of educational products available from U.S. organizations providing genetic tests or services indicated that many did not include significant information about testing risks and benefits, patient rights, or the intended purpose of the test, while test accuracy information was generally limited. Thus, while helpful, these educational aids could be enhanced (62).

Novel approaches aimed at improving care and integrating genetic services into the community include the development of a preconception clinic in the Netherlands, under the joint direction of a gynecologist and a geneticist. The clinic was advertised to PCPs as well as directly to the public. Response was very positive, with many patients indicating they would not have approached their PCP with their concerns, and participating providers noting that many serious issues were addressed through the clinic. In countries where many pregnancies are unplanned, such initiatives may be less applicable, although they may also serve to educate the public about the importance of pregnancy planning (63). Certainly before an expansion of genetic services within primary care can be effectively implemented, more extensive studies of potential tools and mechanisms must be undertaken.

## A Glimpse of Future Challenges

In the future, it is highly likely that information about genetic variations that each confer a small increase in disease risk will be identified, along with insights into gene-environment interactions. In many cases, complex interactions between several genes and environmental agents will be involved in disease causation. As these variations are discovered, predictive genetic testing may be promoted, although such testing may not immediately provide results that are useful for management or risk assessment for all individuals being tested (5). Such complex issues require careful handling by all providers. As genetic tests and preventive therapies are optimized, population-wide screening is likely to increase in scope, a genetic application that will obviously involve PCPs (64). The commercial drive by the pharmaceutical industry to discover the biological explanations that underlie individual variation in drug response could potentially have a significant impact on primary care in the future as well (3).

While PCPs currently give limited attention to genetics overall and some do not yet perceive genetic education as relevant to their practice, increased public demand for genetic advice and the development of genetic tests and treatments of direct clinical value will gradually drive the acquisition of genetic skills. Although the exact future role of non-genetic healthcare providers in genetic service delivery is unknown, it seems certain that some changes in their medical practice will be required. At minimum, improvements in the ascertainment of genetic risk through family history assessment are likely to be necessary to facilitate appropriate referral for more extensive genetic care. Innovative tools, including pedigree-drawing programs, management guidelines, and decision supports, are needed to incorporate these changes. Undoubtedly, online resources will become increasingly important for providers and patients (3). Partnerships between genetic providers and other specialists will also be essential to facilitating appropriate diagnostic, counseling, and management services for a growing number of patients as more genes involved in common diseases are identified.

# 8. THE VIEW FROM OUTSIDE: RESULTS OF THE PRIMARY CARE PROVIDER SURVEY

As discussed at length in the previous chapter, a significant amount of data exists regarding the genetic knowledge, attitudes, and practice patterns of PCPs and other non-genetic healthcare providers across the country. While these studies provide a wealth of data that is helpful in planning for genetic services and education in Virginia, very little is known about the specific genetic needs and attitudes of Virginia's PCPs. Two pilot projects involving non-genetic providers in Virginia provide some information regarding these issues, however. A 2002-2003 survey of urban obstetrician-gynecologists in the Richmond area and rural prenatal care providers in south-central Virginia revealed that while most offer genetic screening tests during pregnancy and refer patients to genetic providers as needed, this occurs less often among rural providers. The vast majority reported educating patients about folic acid, often preconceptually, but more than a third were not offering cystic fibrosis carrier screening as recommended by ACOG and ACMG. The development of referral guidelines, better service accessibility, and increased educational efforts were cited as potential improvement mechanisms for genetic services and referrals. In addition, 67% of rural and 33% of urban providers indicated an inability to stay updated on prenatal genetic issues (1). A separate study of Virginia nephrologists and urologists surveyed their genetic knowledge and practice patterns regarding polycystic kidney disease, a fairly common condition seen within those specialties that is often of genetic etiology. Results revealed that 70-80% of respondents rarely or never refer patients to genetic providers, although they report providing limited genetic counseling themselves. Many do not recommend screening for at-risk relatives or pregnancies and 20-30% incorrectly assessed recurrence risk, suggesting significant gaps in genetic care for these patients (2).

## Primary Care Provider Survey

To obtain more specific data about the needs and attitudes of Virginia's PCPs regarding genetic services and education, the GeneSEAN needs assessment process included the development of a survey addressing these issues that was distributed to PCPs across the state. The survey, which is included as Appendix B, contains questions that address practice patterns and service provision, as well as providers' perceptions of a variety of factors, including the quality and accessibility of genetic services in Virginia, the role of state organizations, and educational needs and practices. Following approval by the VCU IRB, the survey was mailed to 5726 PCPs, including all family practitioners, internists, pediatricians, obstetrician-gynecologists, and VDH district directors currently in practice in Virginia, as identified by the Medical Society of Virginia

A total of 262 completed surveys were returned, for an overall response rate of 4.6%. Within the targeted medical specialties, obstetrician-gynecologists and pediatricians responded at a slightly higher rate, while internists had a lower response rate, perhaps reflecting the fact that internists traditionally interact with genetic services less often than the other specialties. Nevertheless, each of the targeted medical specialties was well represented among respondents. Unfortunately, no demographic data is available regarding non-responders other than medical specialty, thus it is impossible to speculate further regarding the potential differences between survey responders and non-responders. Although the response rate was low, the demographic data reveals that the physicians who responded represent a variety of backgrounds. Males and females were nearly equally represented, while respondents reported a variety of work settings, with 21% rural, 17% urban, and 62% suburban. While the majority of survey participants were Caucasian, other ethnic groups were represented, as shown in Figure 8-1. Years of experience varied tremendously among survey respondents, as shown in Figure 8-2. Overall, it appears that the survey participants represent a broad cross-section of Virginia's PCPs and; therefore, provide a valuable source of insight

regarding genetic services and education needs in the Commonwealth.

The survey results indicate that the majority of Virginia's PCPs are satisfied with the genetic services available in the state. However, many cite gaps in service accessibility in some areas of the Commonwealth, as well as prohibitive costs for some patients. Providers identified genetic education for themselves and the public, the translation of genetic discoveries into clinical applications, and patient education resources as their greatest needs in genetics. In particular, education about practice guidelines--including high-risk patient screening--and genetics referral guidelines were requested by survey participants. Further details about these and other survey findings are discussed below.

**Figure 8-1. Participant Race/Ethnicity**

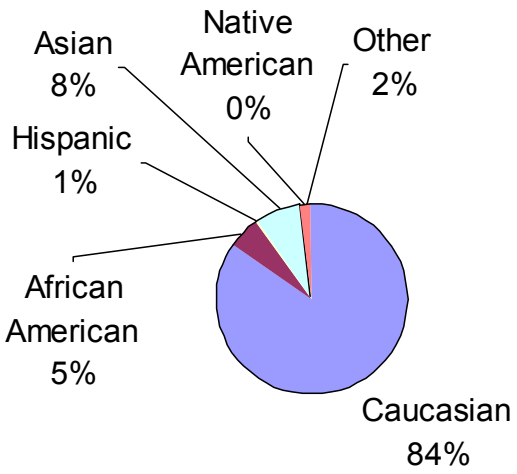
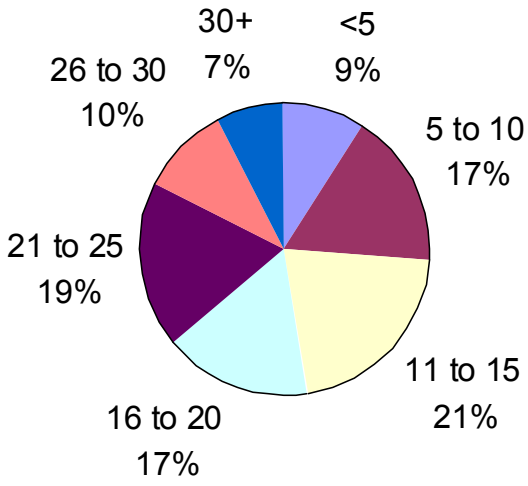



**Figure 8-2. Participant Years of Experience**

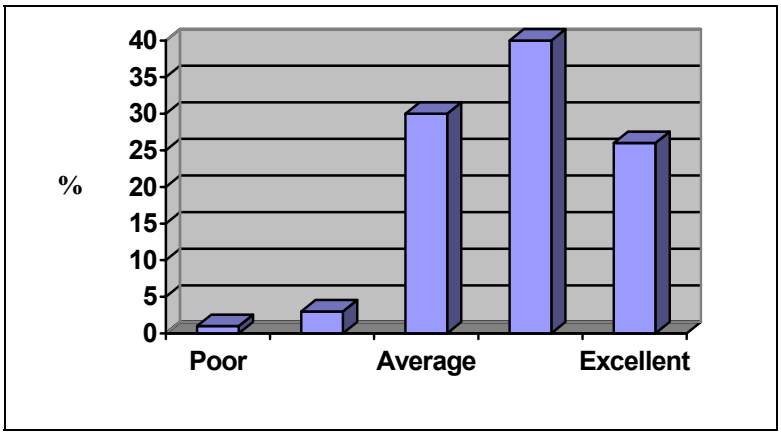



## An Assessment of Virginia's Genetic Services

Overall, Virginia's PCPs report being satisfied with the genetics services available in Virginia, with 66% rating the services as above average or excellent, as shown in Figure 8-3. However, survey participants

did identify some gaps in available services, with 29% reporting that there are not enough genetics services in Virginia. In addition, 25% reported that the cost of a genetics appointment has prevented patients they referred from receiving services, and 22% indicated that distance to a genetics clinic has prevented their patients from obtaining care. The number of referred patients that are not seen for genetics consultation appears to be quite significant, with 18% of PCPs reporting that 50% or less of their referred patients are actually seen by genetics. While cost or distance to a genetics clinic undoubtedly accounts for some of this, there may be other factors that also contribute to this failure to obtain genetic care.

**Figure 8-3. Quality of Genetic Services**




Survey participants were also asked to rank the top three needs in genetics for Virginians. The most common responses were: (1) more genetic education, (2) more application of genetic research to clinical problems, and (3) more educational materials for families. These responses closely mirror needs cited by Virginia's genetic providers and genetic consumers, providing a clear indication of where resources should be directed in the future.

The level of involvement of state organizations and public health leaders in clinical genetic services varies from state to state, with different administrative arrangements supporting the various levels of the public health pyramid from infrastructure support to direct services. When asked to rank the various roles of VDH relevant to genetics in order of importance, Virginia's PCPs appeared to most value the assurance role played by VDH, with participants ranking newborn screening follow up, ensuring availability of genetic services, and ensuring access to lab testing as the most important functions. The role of VDH in genetics education, the birth defects registry, planning for the future in state genetics services, and quality assurance were viewed as somewhat less important by Virginia's PCPs.

### Relevant Practice Patterns

Survey participants reported seeing an average of 3869 patients each year, yet the overwhelming majority indicated that they refer 20 or fewer patients to genetics each year, which amounts to significantly less than 1% of the patients being seen by PCPs. Based on the population incidence of birth defects, genetic disorders, and other health issues relevant to genetics, this suggests that many patients that could be helped by genetic services are not being referred by their PCPs. Improved education of PCPs as well as broader access to services may help address this under-referral problem.

Physicians were also asked how they identify family history of a genetic condition in their practice. Their responses reveal that Virginia's PCPs use a variety of techniques to identify at-risk patients. Most

commonly, PCPs ask patients about their family history to identify conditions for which they may be at risk, with 79% reporting that they use this modality. In addition, 50% reported that patients fill out standardized family history forms at the initial visit, 41% indicated that they ask patients about family history if they express a concern about it, 56% ask patients about family history if it is relevant to their presenting medical condition, 14% refer family history questions to genetic providers, and 1% said they rarely ask patients about family history. When asked how they usually address patient questions about genetic issues, 56% reported that they handle most of these questions within their practice, 40% indicated that they refer most of these patients to genetic providers, and 8% refer most of these patients to specialists other than genetic providers. Several studies have shown that many PCPs and other non-genetic healthcare providers are inconsistent in their assessment and handling of family history issues, thus this is a topic that bears watching in Virginia as well, and one that should be a target of future educational initiatives.

The survey results also indicate that gaps remain in folic acid education practice patterns among Virginia's PCPs. While many survey participants reported discussing folic acid with various patient populations, 20% indicated that they do not discuss folic acid at all. Of those who do address folic acid issues, 62% reported discussing folic acid with women who are planning a pregnancy, 54% discuss folic acid with women who are pregnant, 40% discuss folic acid with women of childbearing age, 12% discuss folic acid with teenagers, and 13% have pamphlets available for patients.

### Genetic Education Needs

Multiple groups—including Virginia's genetic providers, PCPs, and genetic consumers—have identified genetic education as a major need as genetics becomes more integral to the practice of medicine as a whole. When asked to rate their own knowledge of genetics, Virginia's PCPs overall rated their knowledge as average, with a score of 5 on a scale from 1 to 10 with 1 being the lowest and 10 being the highest. Overall, they rated their personal need for genetic education as slightly above average, with a score of 6, while the need for genetic education among physicians in general was ranked slightly higher, with a score of 7. The survey participants indicated that the most important topics to target for genetic education initiatives include cancer, metabolic disorders and other disorders identified by newborn screening, cystic fibrosis, hemoglobinopathies, mental retardation/developmental delay, and birth defects. Virginia's PCPs also reported a need for education on the clinical applications of genetic technologies, including more education on available genetic tests and screening guidelines for patients identified as having a hereditary predisposition to cancer or other diseases. In addition, physicians reported a need for genetic referral guidelines. The most popular genetic education modalities chosen by survey participants were (1) local education/seminars, (2) statewide CME conferences, and (3) journals.

Virginia's PCPs also identified the public as being in significant need of genetic education, with an overall score of 7. The topics most commonly suggested for public education initiatives included cancer, cystic fibrosis, hemoglobinopathies, Down syndrome/trisomies, birth defects, neural tube defects and folic acid prevention, newborn screening, cardiovascular disease, and diabetes. To reach the public, physicians recommended using television commercials and newspaper articles.

## 9. PUBLIC AND PROFESSIONAL GENETIC EDUCATION: EFFORTS AND OUTCOMES

*As the radius of knowledge gets longer,*
*the circumference of the unknown expands even more.*
*~ Anonymous (1)*

A genetic revolution in medicine will require a transformation of genetics education as well, focused on the development of a genetic literacy that facilitates collaboration between patients and health care providers as partners in the promotion of health and the prevention of disease. The most efficacious application of genetics in medicine requires educated patients receiving care from educated providers (2). Francis Collins, director of NHGRI, has provided a hypothetical future clinical scenario, given below, that illustrates the potential application of genetics into the everyday practice of medicine, and highlights the need for public and professional genetic education (3).

***A Hypothetical Future Case***

*John, a 23-year-old college graduate, is referred to his physician because a serum cholesterol level of 255 mg/dl was detected in the course of a medical examination required for employment. He is in good health but has smoked one pack of cigarettes per day for six years. Aided by an interactive computer program that takes John's family history, his physician notes that there is a strong paternal history of myocardial infarction and that John's father died at the age of 48 years*

*To obtain more precise information about his risks of contracting coronary artery disease and other illnesses in the future, John agrees to consider a battery of genetic tests. After working through an interactive computer program that explains the benefits and risks of such tests, John agrees, signing an informed consent form, to undergo 15 genetic tests that provide risk information for illnesses for which preventive strategies are available. He decides against an additional 10 tests involving disorders for which no clinically validated preventive interventions are yet available.*

*A cheek-swab DNA specimen is sent off for testing, and the results are returned in one week. John's subsequent counseling session with the physician and a genetic nurse specialist focuses on the conditions for which his risk differs substantially (by a factor of more than two) from that of the general population. Like most patients, John is interested in both his relative risk and his absolute risk.*

*John is pleased to learn that genetic testing does not always give bad news – his risks of contracting prostate cancer and Alzheimer's disease are reduced because he carries low-risk variants of the several genes known to contribute to these illnesses. But John is sobered by the evidence of his increased risks of contracting coronary artery disease, colon cancer, and lung cancer. Confronted with the reality of his own genetic data, he arrives at that crucial "teachable moment" when a lifelong change in health-related behavior, focused on reducing specific risks, is possible. And there is much to offer. By this time, the field of pharmacogenomics has blossomed, and a prophylactic drug regimen based on the knowledge of John's personal genetic data can be precisely prescribed to reduce his cholesterol level and the risk of coronary artery disease to normal levels. His risk of colon cancer can be addressed by starting a program of annual colonoscopy at the age of 45, which in his situation is a very cost-effective way to avoid colon cancer. His substantial risk of contracting lung cancer provides the key motivation for him to join a support group of persons at genetically high risk for serious complications of smoking, and he successfully kicks the habit.*

While this vision of a future genetic-based, individualized preventive medicine is exciting, its realization requires significant changes to the current health care system, not least of which is a vastly improved knowledge of genetics among health care providers and the general public. This hypothetical scenario is also quite simplistic, while the reality is likely to involve more complicated test result interpretation and clinical management issues. A better-informed public served by knowledgeable providers could significantly improve the current provision of genetic care, in addition to laying the foundation for future changes and expansions in clinical genetic medicine.

### What are the Genetic Education Needs?

As reviewed in chapter seven, significant gaps in the knowledge and practice of genetics by PCPs and non-genetic specialists have been documented in multiple studies. Most assessments suggest a particular need to improve providers' general knowledge of clinical genetics, as well as to increase their risk assessment and referral decision-making skills (4). Surveys indicate that most providers are uncertain about their understanding of genetics and would like more education, with many suggesting the development of referral guidelines as well as educational programs through a variety of modalities (5). Recent medical school graduates and physicians that have regular interaction with genetic professionals have been shown to be somewhat more knowledgeable about genetics, suggesting that courses and increased interactions of genetic professionals with other medical specialties may facilitate improvements in knowledge and practice (6, 7). Most surveys of allied health practitioners show even more critical educational needs, with these groups reporting minimal to non-existent training in genetics despite practice needs (8, 9, 10).

The needs for genetic education of the general public are less well documented, but the available data suggest that the public has only a basic understanding of genetics, with some misconceptions as well, despite what seems to be an almost continuous media focus on genetics (11). Some studies have noted that individuals are generally aware of their limited knowledge of genetics and use a variety of information sources. Many of the studies available addressing public knowledge are actually based on samples of genetic patients or their families, not the general public, with results indicating that consumers have difficulty understanding numerical risks, they often view genetic risks differently than health professionals, and they frequently have different priorities than genetic providers (12). While the results of these studies are useful and reinforce the need to assess consumer knowledge and attitudes about genetics, they are not necessarily representative of the diversity of public knowledge and attitudes. An overview is provided here; however, a much more thorough review of the available information about the genetic knowledge of the American public is available in "Genetics Literacy Project - Literature and Materials Review: A Working Report" (13).

Family members of individuals who have been diagnosed with a Mendelian genetic condition are frequently knowledgeable about genetics, but the general public often is not, nor are patients or family members of individuals with non-Mendelian genetic conditions (14, 15, 16). Many of the studies of public knowledge of genetics have assessed knowledge of the genetics of cancer and other common adult-onset disorders, because these are expected to increasingly be a part of medical care. A Seattle survey of women in a primary care waiting room revealed that the vast majority had heard little to nothing about genetic susceptibility testing and most did not know the answers to questions assessing knowledge of breast cancer genetics. Despite this lack of knowledge, the vast majority expressed interest in genetic testing for breast cancer, and 96% wanted to learn more about it. Respondents indicated a preference for PCPs and informational booklets as information sources (17). Two studies of public interest in breast cancer genetic testing revealed that while many were interested in testing, respondents, including those with breast cancer, showed a significant lack of understanding of the limitations and implications of

genetic testing (18, 19). Another study involving relatives of individuals with colon cancer demonstrated significant misconceptions about what is involved in genetic risk assessment and education, suggesting a need for improved communication with health care providers prior to genetics referral (20). Variable levels of genetic knowledge among different ethnic groups have been noted, as exemplified by an Alabama study of women's knowledge of breast cancer and cancer genetics. Results indicated that African-American women had significantly lower knowledge levels than Caucasian women, despite equivalent income and education levels (21). Variations in knowledge among different high-risk ethnic populations have also been demonstrated, with Jewish Americans showing a much higher level of knowledge about genetic disorders common in that population than Italian Americans did regarding thalassemia, for example (22, 23). For individuals undergoing genetic testing or other procedures, a certain amount of knowledge is a requirement of informed consent, yet this does not always occur, particularly in population screening programs such as obstetric ultrasound. A study of women undergoing prenatal ultrasound revealed that almost half were not aware of its use as a screening test for birth defects, while many did not recall having been informed about the purpose, benefits, limitations, or consequences of testing (24). Interestingly, despite this fairly uniform lack of public knowledge across studies, several have demonstrated a more sophisticated understanding of and concern about the social and ethical issues involved with genetic testing (11, 12). Unfortunately, little improvement in public knowledge of genetics appears to be occurring over time, as exemplified by a Washington state study comparing public knowledge and attitudes in 2003 compared to 1993, with results showing little change apart from a much more widespread awareness of the HGP (25).

The public appears to have a number of misconceptions about genetics as well, including both negative and positive exaggerations (26). Lay conceptions of inheritance are sometimes based on resemblance, with an assumption that physical appearance, personality traits, and disease susceptibility are jointly inherited (27). Multiple studies have shown that the public frequently has an inaccurate perception of disease risk, which may have a significant impact on preventive behavior. A study of individuals involved in a family-based screening program for familial hypercholesterolemia (FH) revealed that participants usually underestimated their risk of having FH and myocardial infarction (28). In contrast, multiple studies have demonstrated that women frequently overestimate their risk of breast cancer (19). Interestingly, while genetic counseling improves the accuracy of patients' risk perception, some studies have demonstrated a persistence of inaccurate self-assessments of risk, thought to result from deep-seated beliefs influenced by the occurrence of breast cancer in the family and by the decisions of other family members at increased risk (27).

Several studies have assessed public attitudes about genetics, with results indicating that most view genetic advances favorably, particularly medical applications, although many are concerned about ethical issues, while other studies suggest a growing suspicion of biotechnology (12, 29). However, variations in attitude have been noted among minority groups. Consistent with the finding that African Americans have a higher level of mistrust of the medical establishment in general, some studies of African Americans show a slightly higher negative attitude toward genetic screening programs than other ethnic groups (30). Interestingly, one study of women in the Seattle area indicated that women with the lowest educational levels were most interested in genetic testing, consistent with other studies that have suggested that such groups are the least informed yet the most willing to accept routine medical procedures (19). Public surveys also indicate that the public believes that they should have access to predictive genetic testing for themselves and their children, regardless of treatment availability (31). Some in the medical community are concerned that a growing reliance on genetic information and testing will result in an attitude of genetic determinism in the patient population, thwarting health behavior modification programs (32, 33). A 2001 study in Georgia suggests that this is not likely to be the case for most Americans, with the majority interpreting genetic status as one of several factors contributing to disease. Again, however, there were variations among different ethnic groups,

with African-American participants espousing a more fatalistic view of genetic status than European Americans (34). Such results provide insights that may be helpful in the design of future educational programs.

Studies of public information sources indicate that the most common sources include health care professionals, the media, and educational settings. Unfortunately, these information sources may not always be reliable. As discussed previously, health care professionals overall have a limited understanding of genetics. The media, which frequently highlights genetic topics, tends to focus on new genetic technologies that often are far removed from clinical application, while the Internet is a largely unregulated information source. A study in Georgia indicated that the public is aware of the drawbacks of the Internet, with participants recognizing the great potential of the Internet for health information on genetics, but expressing concerns about privacy issues as well as the credibility and accuracy of online information (35).

With the discovery of the role of folic acid in birth defect prevention, widespread educational campaigns were initiated to inform the public in the hopes of increasing awareness and use of folic acid, and thereby decreasing the incidence of neural tube defects and other associated birth defects. Results have been positive, although not universal. MOD surveys indicate that by 2005, 84% of women had heard of folic acid compared to 52% in 1995, although only 25% reported knowing that it helps to prevent birth defects. By 2005, 33% of non-pregnant women reported taking a multivitamin containing folic acid daily. Higher income and education levels were positively associated with folic acid use. Of those who were aware of folic acid, the majority had heard about it from media sources, while 25% reported their health care provider as the information source (36). A Canadian study revealed that women who had received folic acid information from multiple sources were much more likely to demonstrate optimal folic acid use before and during pregnancy, suggesting that multi-focused educational efforts are more likely to be successful (37).

Very few studies have specifically addressed genetic knowledge among Virginians. A pilot study in Richmond identified information sources used by the general public, with results indicating that the majority has obtained information about genetics from the media, science courses, and the Internet in the past. Respondents indicated that they would use the same sources in the future, along with health care providers. The study also revealed that the public's interest in genetics largely revolves around health topics, and the vast majority considers an understanding of genetics to be very important. Although respondents' genetic knowledge was not tested, 74% ranked their own knowledge of genetics as average or below (38).

Little data is available regarding Virginians' attitudes toward genetics; however, the results of a November 2003 survey sponsored by the Virginia Advisory Committee on Ethical, Legal, and Social Issues in Genetic Research were recently published, providing insight into the topic. This telephone survey of 804 adult Virginians largely focused on attitudes toward genetic testing as well as genetic privacy and discrimination issues. Results indicated that most Virginians support genetic testing, with 89% saying that genetic testing for the risk of disease is a very good or a good idea. Privacy concerns were more important than any other factor influencing personal decisions to get genetic testing, with the majority of respondents indicating that they were very concerned about their ability to keep health and medical information in general private. When asked about genetic discrimination, the vast majority of respondents thought that life insurance and health insurance companies would deny coverage based on an individual's genetic testing results, while slightly fewer felt that employers would discriminate based on such information. Despite these obvious concerns about genetic privacy and discrimination, 63% of respondents indicated that the benefits of genetic testing outweigh the risks (39).

## Overview of Genetic Curricula in Virginia

The Virginia Department of Education has issued *Standards of Learning* (SOL*)* for public schools in the state, describing expectations for student learning and achievement in grades K-12, including a curriculum framework detailing the specific knowledge and skills required to meet the standards for certain subjects such as science. These SOLs are reviewed and revised at least once every seven years and provide an outline of subject matter included on the standardized assessments required for graduation. Genetics is introduced in middle school as part of the SOL Life Science curriculum, which provides a general overview of common genetic terms and basic principles of genetic science, including the concepts of mutation and the inheritance of traits. Genetics education continues in high school as part of the SOL Biology curriculum. This curriculum plan includes a much more extensive look at the mechanisms of genetics and inheritance, as well as an introduction to the clinical applications of genetics in disease and health, although currently some important topics such as folic acid education are not required elements (40). The Virginia Council on Folic Acid has developed folic acid curricula for incorporation into the SOL, but this has not been adopted statewide (41). For much of the public, this exposure to genetics in secondary school is the sole source of genetics education, other than issues presented by the media. Among adults in the current population, the understanding of genetics may actually be lower, since previous curricula often did not include as much genetic content. Most colleges and universities offer courses in genetics; however, these are rarely part of the required curriculum except for some science majors.

A review of a sampling of health professional training programs in Virginia revealed very limited student exposure to genetic principles. Two of the three Virginia medical schools include dedicated genetics courses as part of their core curricula, with "Medical and Molecular Genetics" at UVA and "Human Genetics" at VCU required during the first year (42, 43). The curriculum at EVMS does not include a dedicated genetics course, although lectures regarding genetic principles are included in other required courses (44). The medical schools also offer several clerkships in genetics; however, these are all elective. Interestingly, the VCU School of Dentistry, which has a long history of genetics involvement, includes "Dental Genetics" and "Advanced Dental Genetics" in its core curriculum (45). Unfortunately, the reviewed curricula of other allied health professional schools in Virginia—including training programs in nursing, pharmacy, physician assisting, physical therapy, occupational therapy, and public health—revealed none requiring a course in genetics, with the obvious exception of the master's program in genetic counseling at VCU. In most cases, these training programs require prerequisites, such as biology courses that often include exposure to basic genetic science, and in some cases, individual lectures on genetics are included as a part of required courses, as in the Master of Public Health program at VCU, where a lecture on genetics and public health was initiated for the 2003-2004 school year. Nevertheless, the students' exposure to clinical genetic principles appears to be quite limited or nonexistent in many instances (46-57).

In contrast, the guidelines issued by national organizations regarding core competencies in genetics for health care providers are quite ambitious. The National Coalition for Health Professional Education in Genetics (NCHPEG), led by the NHGRI, the AMA, and the American Nurses Association (ANA), with over 160 other member organizations, endorsed core competencies for all health care professionals in February 2000. The guidelines provide a minimum level of genetic knowledge, skills, and attitudes necessary for the effective and responsible integration of genetics into the current practice of all health care disciplines, with essential competencies including (1) an appreciation of the limitation of one's genetic expertise, (2) an understanding of the social and psychological implications of genetic services, and (3) a knowledge of how and when to make a referral to a genetics professional. The complete document is provided in Appendix C and can be found on the NCHPEG Web site (www.nchpeg.org). The

guidelines are designed to be used by groups responsible for continuing education, curriculum development, licensing, certification, and accreditation bodies for all health care disciplines, with modifications as appropriate for a particular discipline (58). By late 2001, NCHPEG reported that about 20 professional organizations and educational institutions had used the guidelines to design workshops, courses, and curricula (59). The Association of Professors of Human and Medical Genetics (APHMG) and ASHG have developed guidelines regarding genetic core curriculum and clinical objectives for undergraduate medical students, while the American Academy of Family Physicians (AAFP) has endorsed genetic core competency guidelines for family practice residencies (60, 60, 62). These documents are also included in Appendix C. Genetic curriculum guidelines for continuing and entry-level nursing education programs have also been developed (63).

### Professional Genetic Education Initiatives

A multitude of genetic education programs have been developed for many types of health care professionals; several of the national initiatives are reviewed here, along with examples of smaller projects that have proved successful in other areas. Where educational needs are identified, many of these programs and tools could serve as models for future educational initiatives in Virginia.

As described above, NCHPEG provides a cohesive focus for interdisciplinary, collaborative, and national efforts to promote health professional education about the application of genetic information throughout all aspects of health services (64). In addition to developing the core competencies in genetics, several other projects led by NCHPEG are underway, including the development of an educational CD-ROM for primary care and public health providers that focuses on the genetics of common disease. This project, which will be distributed to all NCHPEG member organizations as well as other interested groups, incorporates two core concepts: that all diseases have a genetic component and that family history is central to a genetically based approach to health care. A second larger NCHPEG program involves the development of six complementary initiatives: (1) *Genetic Resources on the Web (GROW),* which is expected to eventually serve as a primary resource and search engine for high quality genetic information; (2) *NCHPEG Membership Committee*, responsible for expanding the number of groups and individuals involved in genetic education efforts; (3) *Diversity and Cultural Competence Working Group*, focused on improving genetics-related health outcomes for minority and underserved populations; (4) *Family History Working Group*, involved with educational efforts centered on the role of the genetic family history in health care; (5) *Content and Instruction Working Group,* responsible for collecting and reviewing existing educational resources; and (6) *Targeted Education Initiatives*, involved with developing and disseminating education projects targeting different providers each year, including dental providers in 2003 (65).

Undergraduate medical education has been the focus of a variety of genetic education programs, and many genetic courses in Virginia medical schools and elsewhere attempt to provide interesting, clinically relevant experiences (66). An initiative in the U.K. incorporating a medical humanities approach has successfully conveyed to students an understanding of how genetic disorders impact the lives of patients and families against a historical context. The program emphasizes the lived experience of genetic illness and encourages the development of critical reading skills in relation to popular press accounts of genetics by having individual student "reporters" interview volunteers with genetic conditions in their own homes, in addition to group discussion of a number of articles with contrasting stances on ethical issues in genetics (67). A project at Harvard Medical School has attempted to expand students' exposure to genetics into the clinical curricula by including genetic specialty clinic participation and an exercise involving the solicitation of a family history on all inpatient admissions during the third year clerkship in medicine, in addition to the incorporation of clinical case discussion into the first year genetic course (68).

In anticipation of a growing need, a group in Israel collected learning resources in pharmacogenetics as part of the development of a pharmacogenetics course targeting undergraduate and graduate students that was later adapted for the medical school curriculum. Future plans include modification into a continuing education module (69).

PCPs are arguably the target of more genetic education initiatives than any other group at present, consistent with the expectation that they will be responsible for providing an ever increasing amount of genetic services despite significant gaps in current knowledge and awareness. The *Genetics in Primary Care (GPC)* project is a national initiative focused on developing primary care faculty expertise in genetics, with the ultimate goal of enhancing the training of medical students and primary care residents. The key goals of the curriculum are to improve PCPs' ability to (1) consider inherited disease in the differential diagnosis of common disorders; (2) use appropriate counseling strategies for genetic testing and diagnosis; (3) and understand the implications of a genetic diagnosis for family members (70). The curriculum includes links to relevant Web sites, articles from the medical literature, and consensus and policy statements, and is available online at http://genes-r-us.uthscsa.edu/resources/genetics/primary_care.htm (71). The Genetics Interdisciplinary Faculty Training (GIFT) program is a similar initiative based at Duke University that targets nurse practitioner, nurse midwife, and physician assistant faculty (72). Surveys of PCPs have indicated a preference for various educational modalities (73). A preference for computer-based resources led a California group to develop a cancer genetics Web site aimed at facilitating appropriate identification, intervention, and referral of patients at risk for hereditary non-polyposis colon cancer (HNPCC) by PCPs (74). Another California group has developed a cancer genetics curriculum for PCPs involving individual lectures and day-long conferences that has been successfully used with 7400 health care providers of various backgrounds (75). Other projects have specifically targeted PCPs in community settings that often have limited access to continuing education, while a variety of tools have been developed that help facilitate the implementation of new practice guidelines, such as cystic fibrosis screening, into primary care practice (76, 77, 78).

A variety of projects targeting different medical specialties have been developed as well. Programs include pharmacogenomics for pharmacists, an educational campaign developed by the American College of Cardiology for its members, and an annual "Genetics Summer Institute" for Ohio nursing faculty (79, 80, 81). A number of pharmaceutical companies have also developed genetic education programs. The ACMG, NSGC, and the Endocrine Society developed an interactive educational exercise for implementation at the annual meeting of the Endocrine Society that involved assigning members a genetic condition and genetic test results, with subsequent genetic counseling experienced first-hand by participants. Future plans include modification of the exercise for use at other specialty meetings (82). As described by the Institute of Medicine Report on the Future of Public Health, public health agencies will have an increasing role in assessing the health needs of populations; working with the private sector in ensuring the quality of genetic tests and services; and evaluating the impact of interventions on medical, behavioral, and psychosocial outcomes (64). This growing public health role is exemplified by a 2001 CDC workshop involving a wide range of stakeholders that identified public health strategies useful for the improvement of health outcomes in individuals with primary immunodeficiency diseases (83). Several initiatives have been developed to enhance the genetic education of individuals involved in public health, including an Ohio project targeting allied health providers in public health that involved an inservice on the benefits of folic acid along with the provision of tools for patient education (84). In addition, the Association of State and Territorial Health Officials (ASTHO) has undertaken the *Genomics Toolkit* project to assist health agencies with integrating genomics into public health practice and policy, while the Universities of Michigan and Washington have each developed graduate training programs in public health genetics, and the CDC offers several training programs in public health genetics, from a short introductory course to a 3-year career development opportunity for public health professionals in genetics (85, 86).

Web-based resources have emerged as particularly convenient tools for providers and the public. Several Web sites, including *OMIM, GeneTests,* and *GeneClinics,* have served as critical information sources for genetic providers for a number of years, and are increasingly used by other healthcare professionals, patients, educators, policy makers, and the media, while Web sites maintained by the MOD and others are frequently used for patient education. *GeneTests* and *GeneClinics* serve as a database of available tests for genetic conditions, a directory of genetic laboratories and clinics, and a resource providing information about the diagnosis, management, and genetic counseling of specific inherited diseases (87). *OMIM* is a comprehensive, authoritative database of human genes and genetic conditions compiled to support the practice of clinical genetics as well as research and education in human genomics (88). Reviews of online resources for genetic providers, PCPs, and genetic patients in the U.S. and the U.K. found several gaps, including a dearth of high quality resources for PCPs. Sites recommended by the U.S. study are listed in Table 9-1, while recommendations for future development included the development of resources containing summaries of genetic screening protocols as well as a standardized "pedigree tool" that efficiently combines population data with family history information to identify high risk patients (89, 90).

**Table 9-1. Recommended Genetic Web Sites Evaluated Using Mitretek Criteria (89)**

| | |
|---|---|
| **For genetic professionals:** | |
| OMIM | http://www.ncbi.nlm.nih.gov/Omim |
| GeneClinics | http://www.geneclinics.org |
| GeneTests | http://www.genetests.org |
| Kennedy Institute of Ethics | http://kennedyinstitute.georgetown.edu/index.htm |
| NNSGRC | http://genes-r-us.uthscsa.edu/resources/newborn/state.htm |
| National Center for Hearing Assessment and Management | http://www.infanthearing.org |
| **For primary care providers:** | |
| National Cancer Institute | http://www.cancer.gov/ |
| **For patients:** | |
| Genetic Alliance | http://www.geneticalliance.org |
| Genetic Education Center | http://www.kumc.edu/gec/geneinfo.html |
| Family Village | http://familyvillage.wisc.edu/index.htmlx |
| NOAH | http://www.noah-health.org |
| Genes and Disease | http://www.ncbi.nlm.nih.gov/disease |

The media serves as a significant educational resource for the general public as well as healthcare providers, yet media portrayals can be inaccurate and misleading. Several educational initiatives have attempted to develop providers' critical reading skills regarding popular press coverage of genetics; however, these programs are not widespread. A recent review of provider response to media coverage of genetics revealed that the attitudes of providers reflected the content of the media coverage to which they had been exposed, and that the relevant newspaper and television reports focused more on medical implications while radio reports more often involved ethical issues (91).

Most provider education efforts in Virginia involve in-services, grand rounds, and similar programs within the academic centers that house genetic departments or divisions, as well as the everyday interactions with referring providers that serve as a mechanism for informal education. Some formal genetic education occurs in outreach areas, community practices, and health departments, although this

appears to be somewhat limited currently. Genetic providers are involved in teaching medical and dental students as well as residents at all of the medical and dental schools in Virginia, and have incorporated several innovative projects into the genetic curricula, including assignments that introduce students to online genetic resources and modules involving cancer patients that acquaint students with the intricacies of predictive genetic testing (92, 93). *Infogenetics*, targeting non-genetic providers, is a Web-based tool developed by the genetics group at CHKD that provides links to a variety of online resources that can be useful in dealing with genetic issues, including many of the sites listed in Table 9-1 (94).

### Public Genetic Education Initiatives

Genetic education of the public occurs through many avenues, including science education in secondary schools, colleges, and universities, as well as adult education through a variety of modalities such as healthcare provider interaction, the media, the Internet, public education campaigns, and direct-to-consumer marketing of genetic testing and services. While much of the process of genetic counseling involves the education of patients, this aspect is not addressed here, since genetic patients represent only a small subset of the general public. A consideration of cultural issues is a key factor in the development of successful educational initiatives for the public. In particular, establishing and maintaining trust, respecting healthcare beliefs and practices, and understanding the impact of culture on decision-making processes are critical (95, 96). Several groups have attempted to assess relevant cultural issues for incorporation into educational materials. Two such projects include the *Genetic Education for Native Americans (GENA)* program, which has successfully provided culturally competent education about genetic research to Native American college students and has increased awareness of careers in genetics, and the *Communities of Color and Genetics Policy Project*, which has engaged African-American and Latino groups in dialogues relating to genome research and related technologies (97, 98).

Secondary school education programs can provide a very effective vehicle for widespread public education. Many community-based programs have been developed throughout the country, although some provide more systematic and complete outreach than others do (99). Those involved with genetic education programs in schools have found that successful efforts require the collaborative efforts of science teachers, genetic researchers, genetic healthcare providers, ethicists, and business partners. Two examples of broad-based, comprehensive educational outreach programs by academic genetic centers include the programs of the Department of Genome Sciences at the University of Washington in Seattle (http://www.gs.washington.edu/) and the Genetics Education Center (GEC) at the University of Kansas Medical Center (http://www.kumc.edu/gec/). Current projects in Washington include workshops and seminars for teacher education, including a week-long summer institute, as well as a Web resource that contains genetic curriculum and resource reviews, classroom activities and tools, and listings of other genetic education programs (100). The GEC site provides information and resources about human genetics and the HGP, as well as lesson plans and links to other relevant sites (101, 102). Other organizations involved with K-12 education efforts include the National Laboratories involved with HGP research, which have outreach programs in their local communities. The NHGRI has an Outreach and Education Office that supports several educational activities, including a Web site with a genetics glossary, media events of the day, and a curriculum supplement in genomics aimed at high school students. A number of public museums, including Virginia museums, have had exhibits focused on genetic issues, providing education for all age groups. Some biotechnology companies and health care organizations have funded or directly developed public outreach programs, while professional genetic societies, including ASHG and the American Society for Gene Therapy, have implemented educational programs for high school students and teachers as part of national meetings (100). A variety of innovative exercises have been developed for use in undergraduate genetics courses, including measuring hereditary traits in cats and using online resources to write genetic papers, however these courses reach a much

smaller proportion of the population (103, 104).

Multiple formats have been used to educate adults about genetics, including traditional written materials, which have been shown to be effective at influencing health behaviors, improving understanding of the risks and limitations of genetic testing, and decreasing interest in testing among low-risk individuals, a group that could overwhelm existing genetic provider resources as more tests become available (105, 106). The Genetic Alliance (www. geneticalliance.org), an umbrella organization for genetic support groups, facilitates a variety of public education initiatives as well, while the NHGRI and the Office of Rare Diseases (ORD) within NIH have developed "The Genetic and Rare Diseases Information Center", which offers genetic information by telephone, fax, e-mail, or postal mail (888-205-2311) (107, 108). As part of their consumer health initiatives, the National Library of Medicine has recently developed the *Genetics Home Reference,* designed to make the health implications of the HGP accessible to the public (109). In some cases, curricula targeting adults have been developed by academic groups, such as the *Learning Exchange for Genetic and Environmental Disease Solutions (LEGENDS)* project implemented in Ohio (110). While many resources are available for individuals who actively seek it, passive genetic education, which is likely to reach a larger audience, is not as widespread. Exceptions include folic acid educational initiatives and a New York campaign involving cancer genetics education, which have used multiple media outlets as well as printed materials (111, 112). A folic acid campaign, which included television, radio, and newspaper ads as well as billboards, a news conference, distribution of educational literature in the community, and secondary school and nursing school interventions, was implemented in southwestern Virginia in 1997. Measures of folic acid awareness increased from 31% to 54% immediately following the campaign, and further improved to 77% one year later, possibly due to simultaneous national campaigns (112).

Computer resources have emerged as a preferred educational vehicle for some groups, and a variety of such tools have been developed (113). The NCHPEG Web site lists two CD-ROM resources that target lay audiences, including one available in several different languages (www.nchpeg.org/eduresources/clearinghouse.asp) (93). Interactive computer programs about cancer genetics have been shown to increase knowledge levels as much as genetic counseling, while patients actually prefer computer programs for some components of the educational process because of its ability to be self-paced and private (114, 115). *Access Excellence* is a national educational program that uses the Internet to connect high school biology teachers with colleagues, researchers, and information resources on genetics and other topics, while *Natural History of Genes* and the *DNA Learning Center* at Cold Spring Harbor are Web-based programs that provide information and activities for teaching genetics (100). The Internet contains a wealth of information on genetics, although not all sites are accurate and reliable (116). A study in Colorado and Wyoming revealed that approximately half of families seen in general genetics clinics had used the Internet to find information about genetics prior to their appointment, although only 5% had been referred to a particular site by a physician. Most used the Internet to get information in lay terms, to learn about treatment options, and to get information about research, although many found the information to be confusing and potentially inaccurate. The vast majority wanted information about recommended sites from genetic providers (117). Such insight suggests that genetic providers should be proactive in promoting high quality Web resources for patients and the public.

Media outlets also provide a significant amount of public education in genetics, although there are drawbacks and limitations involved in these as well. Some provide high quality, accurate information with well-rounded perspectives. The documentary "A Question of Genes," produced by the Public Broadcasting Station and available on video with an accompanying teacher's guide, explores the challenges confronted by families at risk for genetic disorders as they consider genetic testing, while the radio series "The DNA files," which focuses on the social implications of genetic research, is available on

tape (100). News stories on genetics are generally "framed" in a particular way, often subtly so, which may influence public perceptions significantly. New genetic discoveries and the medical benefits of genetic research are commonly covered in the news, particularly print news media, while ethical issues and multifactorial interactions of genes and environmental factors are less commonly discussed (118). Direct-to-consumer advertising for genetic testing has also emerged recently, acting as a source of public education as well as influencing public demand for genetic services. Some have argued that such ads are inappropriate, citing the complexity of the information, the social context surrounding genetics, and the lack of consensus about the clinical utility of some genetic tests (119). A study by Kaiser Permanente found that referrals to their clinical genetics facilities in Denver, where Myriad Genetic Laboratories initiated extensive advertising for breast cancer genetic testing in September 2002, increased more than 300% during the campaign compared to the previous year, yet much of the increase consisted of low-risk patients (120). Such exposures help shape the agenda for public debate and can certainly affect public demand for services and are, therefore, important areas of investigation.

# 10. PUBLIC PERCEPTIONS OF GENETICS: RESULTS OF THE CONSUMER FOCUS GROUPS

## Overview

A complete assessment of Virginia's genetic service needs requires input from genetic consumers, thus this needs assessment process included the solicitation of consumer feedback through focus groups of genetic patients throughout the state. Unfortunately, little information regarding the attitudes and opinions of genetic consumers is available in the literature. As discussed in more detail in the previous chapter, several studies have measured the genetic knowledge of consumers, while a few have compared the efficacy of various teaching modalities used with genetic patients. Many more studies have assessed genetic knowledge and attitudes among the general public, as reviewed in detail in chapter nine. In a handful of other cases, pilot projects have measured consumer response to unusual genetic service delivery methods such as telemedicine, as discussed elsewhere in this report. Despite the fact that many medical centers throughout Virginia conduct patient satisfaction studies, there are virtually no data available regarding satisfaction levels among Virginia's genetic consumers specifically. EVMS is currently conducting a study that may provide such specific information; however, no results are yet available (1). To address this relative lack of information about consumers' impressions of genetic services in Virginia, a brief survey and a group process script were developed for use in a focus group setting and implemented in two regions of the state. These tools, along with a variety of related materials developed for the focus group project, are included in Appendix D. Consumer input regarding multiple issues was solicited through the focus groups, including the quality and accessibility of genetic services in Virginia, genetic services needs, educational needs and practices, and genetic discrimination and privacy. The results provide critical insight into current practices and gaps in genetic health services and education that was not available previously.

Once the focus group tools and materials were approved by the VCU IRB, secondary approval was obtained from the UVA and EVMS IRBs, because focus group participants were to be recruited through genetic clinics under the purview of those organizations as well. Genetic providers at CHKD, EVMS, UVA, and VCU facilitated participant solicitation by distributing informational flyers to patients attending prenatal and general/pediatric genetic clinics in Fredericksburg, Lynchburg, and Norfolk, Virginia. Interested patients were asked to telephone GeneSEAN project staff for further information. Individuals who elected to enroll as focus group participants were sent a confirmation letter providing further information about the purpose and format of the focus group sessions. Upon arrival for a focus group session, participants completed an informed consent document and a brief survey. A group discussion moderated by project staff followed, utilizing the moderator script included in Appendix D. The discussions were recorded; however, participants remained anonymous. Upon conclusion of the focus group session, each participant was given a $25 gift certificate. Project staff then reviewed the session transcripts to identify key findings and central themes. Those results, along with the survey data, are summarized below.

Participants for the Fredericksburg focus group session were recruited from prenatal and pediatric genetic clinics staffed by genetic providers from VCU. Four patients responded to the informational flyer; however, only three were able to participate in the focus group session. Lynchburg participants were recruited from a monthly general/pediatric genetic outreach clinic staffed by UVA genetic providers. One patient responded and participated in the focus group meeting. For the Norfolk session, participants were recruited from prenatal genetic clinics held at EVMS and pediatric genetic clinics held at CHKD. Two patients responded; however, the Norfolk focus group session was not able to be held in 2004. Individuals

who chose to participate tended to be highly motivated to share their views on genetic services and, in most cases, had each had multiple interactions with genetic providers and genetic services in Virginia, while at least one participant also had experiences with genetic services in other states. Although it is highly likely that these characteristics do not reflect those of many non-participants, such depth of exposure to genetic services among those who did participate provides a rich source of useful information. Further details about non-participants are not available. The three sites of the focus group sessions were chosen because of their geographic distinctions, with Fredericksburg being fairly suburban, while Lynchburg is in a mainly rural area, and Norfolk is a large urban center. Demographic information about participants shows a range of characteristics as well, despite the small numbers. Three of the four total participants were female, while half were African American and half were Caucasian. Participants' ages ranged from the 20's to the 40's. Educational level also varied, with one participant having a high school diploma or equivalent, one with a bachelor's degree, and two with master's degrees.

Overall, focus group participants were quite satisfied with their interactions with genetic providers in Virginia and felt the quality of services provided was above average. Nevertheless, they did offer multiple suggestions aimed at improving both genetic specialty care as well as the general medical care available to individuals with genetic conditions in Virginia. Participants identified gaps in their own as well as their PCPs' knowledge of genetics and genetic services, and encouraged increased genetic education on multiple levels. None of the focus group participants felt they had experienced genetic discrimination or privacy infringements; however, poor insurance coverage of genetic services was identified by participants as a form of discrimination against individuals with genetic disabilities. Further details regarding the insights gained from the focus groups are discussed below.

### Virginia's Genetic Services: A Quality Assessment

Focus group participants were asked to assess the quality of the genetic services they had received through one question included in the focus group survey as well as several questions raised during the group discussion. Overall, participants' opinions of Virginia's genetic services were positive. On the survey, assessments of service quality varied slightly, with one participant rating Virginia's genetic services as being of average quality, while the other three indicated the quality level was good or excellent. During the group discussions, all participants expressed satisfaction with the genetic services they received in Virginia. Participants also provided insights into aspects of their experiences that were especially helpful. In particular, participants valued the ability of genetic services to address concerns that the PCP could not, the sharing of relevant information with the PCP following the genetic consultation, the provision of up-to-date information and immediate feedback, and the thoroughness of the genetic consultation and exam.

One participant emphasized the clarity and conciseness with which information was provided to him at the time of genetic consultation. Participants also appreciated the amount of time the physicians spent with them in order to explain all of the necessary information during their appointments.

*"I feel that when they are explaining those types of things they really need to take their time so you completely understand the process… and both of them did that."*

*"I think they were really good, I think both doctors really sat down and took a lot of time."*

Participants were impressed by the comprehensive review of their family histories, including not only the history relevant to the referral indication, but the remainder of the family history as well. They also noted the thoroughness of the physical examinations performed when indicated, as well as the inclusion of all

family members, including children, in the consultation.

*"It was nice that she didn't make it all about him and that she was including me and making it a big picture."*

*[Speaking about her three year old daughter] "She (the physician) actually talked to her… I like that she actually tried to communicate with her…"*

Participants also valued preventive and proactive information when obtaining genetic services. They expected an increased level of thoroughness and expertise from genetic providers compared to their other healthcare providers, with a focus on improving their own genetic health and that of their current or future children. Participants appreciated information and educational materials related to maximizing their health during pregnancy as well as ensuring the health of their unborn fetus. One participant also noted that it was important for geneticists to provide practical tips and applications for everyday life with a genetic condition
and professional input about problems that might arise in the future, and how to avoid or minimize their effects.

*"Being proactive is very important."*

*"In seeing a geneticist you expect a greater amount of detail."*

*"…basically…what can we do to be as educated and knowledgeable to deal with it and make it the best situation for the child."*

Another participant appreciated the fact that the genetic consultation provided him with an opportunity to learn about his disability and gain closure on some unanswered questions, particularly the origin of his disability.

*"I think part of doing this…is to find what I have and answer how it happened and try to make those connections and rule out and to see the likelihood of some of the problems we might be facing in having a family."*

Overall, participants reported having no significant difficulties accessing genetic services in Virginia. One participant was unaware that genetic services were available locally and, therefore, traveled to get those services. Another participant reported that a genetic consultation had been postponed due to a delay in the transfer of their medical records.

### Identified Gaps and Improvement Mechanisms

Several questions included in the focus group discussion addressed participants' perspectives on gaps in genetic services and solicited their recommendations for improvement. Participants identified several areas in which genetic services as well as general medical care for individuals with genetic conditions could be improved and offered multiple suggestions for enhancing services. Participants' views and comments are discussed below, while Table 10-1 provides a summary of the methods suggested to improve genetic services.

In some cases, participants indicated that having more information beyond what was discussed during the genetic consultation would have been helpful. Participants expressed a desire to be completely involved and educated about the topic of genetics and the consultation process, including having information about

why particular genetic disorders or syndromes were ruled out by the geneticist. Recommendations for improvement included an increased use of handouts and other resources that provide detailed information about relevant syndromes.

Some focus group participants indicated that they were not fully informed about what would be expected of them during a genetic consultation, while another participant expressed anxiety regarding the reliance on her memory to provide significant historical information and background. All suggested that the provision of instructions or a checklist to facilitate patient preparation prior to scheduled appointments would increase the efficacy and usefulness of the consultation and improve the accuracy of the historical information provided by the patient.

*"It might be a good suggestion to say that when you come to this consult you will need to know x or y and the doctor would say that would be really helpful to us."*

*"I think a more formal checklist of things that may be asked so you can prepare beforehand, I think that would be very useful. It would speed things up and I think the information would be more useful."*

One participant indicated that family history information should be obtained from medical records, with only an abbreviated review at the time of follow-up genetic appointments, so that the consultation time could be devoted to a discussion of current and upcoming patient needs and concerns.

*"I was expecting that the past information from the previous doctor would be transferred and I wouldn't have to do all of that again."*

Another gap identified in the provision of genetic services in Virginia was the lack of coordination of care among different medical specialists involved in the care of individuals with genetic conditions. To ameliorate this need, one participant recommended that genetics centers provide a list of local providers in different specialties that have experience in caring for individuals with various genetic conditions.

Participants also identified a need for more local support groups for specific genetic conditions. They believe that support groups would facilitate connections with others that had been through the same or similar situations and would provide opportunities to gain valuable information drawn from others' real life experiences.

*"A support group would be nice to talk to somebody that has been through this so you can further educate yourself."*

**Table. 10-1: Suggested Service Improvement Mechanisms**

| |
|---|
| ▪ Provide names of physicians that have experience working with specific genetic conditions. |
| ▪ Improve coordination of care between specialists. |
| ▪ Streamline information-sharing between genetic providers and other specialists. |
| ▪ Conduct multidisciplinary meetings with various specialists caring for individuals with genetic conditions. |
| ▪ Provide a questionnaire and instructions prior to initial genetic consultation. |
| ▪ Develop more support groups for various genetic conditions. |
| ▪ Increase early intervention by targeting parents during ob/gyn and pediatric appointments. |
| ▪ Increase government involvement in genetic issues. |
| ▪ Conduct telethons. |
| ▪ Develop camps and workshops to increase community support regarding genetic issues. |

## Genetic Education Needs

Genetic education and awareness needs were addressed by questions in the focus group survey as well as the group discussions. Generally, participants believe genetics was somewhat or very important for society; however, all rated their own genetic knowledge and understanding as being of average level, suggesting that, despite dealing with genetic issues in their own lives, they recognize a gap in their genetic knowledge. Participants also indicated that their PCPs and other non-genetic specialists were often unaware of genetic services and had limited knowledge about genetic conditions; however, they applauded their providers' willingness to provide referrals to specialists able to address genetic issues. Participants also indicated that they were not concerned about their physicians' limited knowledge of genetics; rather they appreciated their honesty about their lack of experience and were impressed with the diligence with which the PCPs located an appropriate referral.

*"He basically said there is not much I can do for you but let me do some research and find out who can."*

Focus group participants advocated an increased involvement of obstetrician-gynecologists in proactively addressing genetic issues and educating patients, particularly regarding preconception genetics. They also identified teachers—particularly those involved in special education—as well as educational administrators as groups in need of education regarding genetic conditions and genetic services.

Regarding information sources, participants reported obtaining information about genetics through physicians, books, and the Internet. One participant reported that once her daughter was born she was given literature about genetics and other relevant information pertaining to her child's condition.

*"Through independent research, books, Internet, things like that, to try to get a handle on, here is what I have and here is the possibility, can it or can't it be passed [on]."*

One participant remarked about the overall lack of public information regarding genetic and disability-related services in Virginia and suggested more widespread production and distribution of materials dealing with the issues of genetics and disabilities. Such increased publicity was viewed as a way to improve the knowledge and awareness of all individuals in the Commonwealth while simultaneously decreasing the false assumptions and stereotypes that can accompany the diagnosis of a disability.

### Genetic Discrimination and Privacy

All of the participants reported feeling satisfied and confident about the level of privacy they perceived while obtaining genetic services, while none seemed concerned that their information would be shared inappropriately or indiscriminately.

*"I think in maintaining confidentiality it is where it needs to be, there is definitely a standard of professionalism there…"*

Although none of the participants reported feeling discriminated against due to obtaining genetic counseling, one participant expressed frustration about the lack of coverage provided by insurance companies and viewed the limited insurance coverage for genetic testing as a form of discrimination.

"Insurance covers a lot of things they shouldn't cover and they don't cover a lot of things; to me genetic services and genetic counseling is a no-brainer, which in a roundabout way is discrimination against me for having a disability."

## 11. EVALUATING THE VIRGINIA SYSTEM: COMPARISONS TO CORN GUIDELINES

CORN, formed in 1985 through funding from the MCHB of the Health Resources and Services Administration (HRSA), provided a forum for dialogue and national coordination among the ten U.S. regional genetic services networks. CORN also played a key role in integrating genetic services into public health programs, particularly maternal and child health programs, performance partnerships, and QA outcome measures. With the explosion of scientific knowledge and technology associated with the HGP, CORN and other national public health organizations recognized the imperative for federal leadership to identify areas of need in genetic services and to provide guidance in the development of new services. While public health programs must plan to cope with the increasing genetic knowledge and technology in order to serve the public well, the genetics community should look to public health systems to translate genetic advances into human services, illustrating the benefit of a partnership between these two groups. With this in mind, CORN and MCHB sponsored a 1996 conference, "Genetic Services: Developing Guidelines for the Public's Health," with the goal of developing a set of guidelines that could be used in planning for the future of genetic services in public health programs. The resulting document provides state and territorial public health agencies with an outline of suggested components for a genetic services system, and is intended to serve as a framework for an assessment of needs within all types of genetic services in a region, in addition to being a guideline for the development of a comprehensive genetic plan (1, 2). Although CORN no longer exists, collaboration between states and national support for public health genetic systems continues through NNSGRC, and regional genetic networks are ongoing with support from a variety of sources.

The complete CORN document "Guidelines for Clinical Genetic Services for the Public's Health," initially published in April 1997, is available on the NNSGRC Web site (http://genes-r-us.uthscsa.edu/resources/pdf/geneticguidelns.pdf). The guidelines facilitate an assessment of the organizational and administrative structure, prevention activities, services availability, research and educational activities, data collection and documentation methods, and funding sources for genetic services within a state or territory, while also addressing genetic staff qualifications, privacy and confidentiality needs, and other issues relevant to the provision of genetic services. Although they provide a framework for the development of a state genetic system, the CORN guidelines are not meant to provide guidance about the practice of clinical genetics. Following publication of the guidelines, a "State Assessment Tool," which facilitates the comparison of a state's genetic services with the CORN guidelines, was developed. A slightly modified version of this tool is used in Table 11-1 to compare Virginia's genetic services to the CORN recommendations. A more comprehensive discussion of the findings is provided below (2, 3).

The CORN guidelines regarding organization call for a state genetics unit or, at minimum, a full-time genetics coordinator with a background in genetics, public health, and service delivery responsible for a variety of activities dealing with advocacy; collaboration and communication; services needs identification; service coordination; consumer involvement, education, and satisfaction; legislative issues; financing of services; and strategic planning; as detailed further in Table 11-2 (2). Virginia does have a dedicated state genetics coordinator, a position traditionally occupied by individuals with backgrounds in nursing and public health. This position involves responsibility for a broad range of activities, including a large administrative component mainly dealing with oversight of newborn screening services, the birth defects registry, contracts with genetic centers, and assurances under Title V with local health departments. While many of the activities recommended by CORN are supported by the Virginia system, some are not, largely due to staffing and funding constraints. Communication with the genetics community is generally limited to the main centers receiving grant support from the state, while other

genetic providers in the community have minimal to no involvement with the state coordinator or the VaGAC; however, efforts to broaden participation are underway. Although data regarding genetic services has been collected from the centers receiving state grants for a number of years, this information has not been thoroughly analyzed. Virginia does have a state genetics advisory committee, VaGAC, which is involved with the activities recommended by the CORN guidelines, with recent initiatives to include consumers on VaGAC and ongoing efforts to develop a state genetic plan. VaGAC also includes a representative of the Virginia Leadership Education in Neurodevelopmental and related Disabilities (LEND) program, as recommended by CORN. This GeneSEAN document represents initial efforts toward the development of a state genetic plan for Virginia. Although details regarding the structure and content of Virginia's state genetic plan are currently undecided, this needs assessment report does describe the state, demographic profiles, genetic systems, relevant data collection systems, QA systems, and funding mechanisms, as recommended in the CORN guidelines regarding the assessment and policy development aspects of a state genetic plan. Efforts will also be made to include the recommended assurance and collaboration activities in the Virginia state genetic plan.

As shown in Table 11-1, a full range of genetic services are available in Virginia, although some areas of the state are better served than others. As described more thoroughly in chapter four, Virginia's clinical genetic facilities are mainly centered in areas of the state with high population densities. While there are some outreach clinics in rural areas, those residents generally have to travel significant distances to obtain services. The Northern Virginia area remains underserved as well. In particular, metabolic and single disease clinics are available in only a few areas of the state. With the exception of newborn screening, birth defect registry enrollment, and early hearing detection and intervention, population-based services are variably available in Virginia, with consumers in areas removed from the main genetic centers less likely to obtain services. While there is no coordinated teratogen information service in Virginia, genetic providers throughout the state do provide teratogen counseling. Adult genetic screening services remain limited. Genetic centers in Virginia generally do meet the CORN recommendations regarding facility and operational requirements, genetic evaluation components, and ethical, legal, and social issues, although active QA and consumer feedback activities are not universal throughout the state. Virginia's genetic professionals generally meet the CORN credentialing recommendations, although the availability of genetic providers is limited in some areas of the state. In addition, a few genetic counselors in Virginia do not meet the training and certification recommendations in the CORN guidelines. Although cytogenetic, biochemical genetic, and molecular genetic laboratory services are available through several labs in Virginia, insurance requirements sometimes result in poorly coordinated care. Some insurance providers require the use of national laboratories, while in other cases, particular genetic tests are not available in-state and require the use of an out-of-state laboratory that may not be covered by insurance. As noted in section III of Table 11-1, funding for genetic services in Virginia comes from a variety of sources, although for many citizens, coverage remains a problem.

The CORN recommendations include a variety of potential sources of data regarding available genetic services and needs, many of which are used in Virginia. The main principle highlighted in the guidelines regarding these sources is the requirement to consider the purpose and primary function of each data source when evaluating its utility for this purpose. Of note, Virginia has no state-level clinical genetics database of clients served, which limits the ability to assess service needs in the state. In addition, the directory of genetic service centers provided by the state genetics unit is limited to those centers that receive state funding and is thus incomplete. There has been a lack of linkages between the various databases relevant to genetics in Virginia, including those the CORN guidelines deem essential for monitoring the occurrence of specific genetic diseases, outcomes in infants and children with those diseases, and assessment of service utilization and efficiency of service delivery; however, major efforts are underway to develop and enhance some of those linkages through the VISITS project and the Virginia

CATPIP grant. The VISITS project involves the development of a Web-based integrated database system that tracks and manages data collected from four programs: the Virginia Early Hearing Detection and Intervention Program, Virginia Newborn Screening Services, VaCARES, and referrals to the At-Risk for Developmental Disabilities Program. The Virginia CATPIP grant involves improvements in the birth defects registry system, including integration of surveillance data with other public health programs.

Although not addressed in Table 11-1, the CORN guidelines also include recommendations regarding prevention, research, and education activities within genetic services. Primary prevention activities include folic acid education, monitoring, and outcome evaluation; teratogen information services and education; appropriate maternal disease management; reproductive genetic counseling; and education and surveillance of individuals at high risk for adult disorders such as cancer. Most of these primary prevention activities are ongoing in Virginia at the local level, while coordinated statewide efforts include the Virginia Folic Acid Council, supported by multiple agencies including the MOD and VDH, as well as birth defect prevention activities within Virginia CATPIP. Recommended secondary prevention activities include newborn screening, prenatal screening, and genetic screening for select high-risk groups, all of which are fairly widespread activities in Virginia. Tertiary prevention activities aimed at ameliorating the unfavorable consequences of existing genetic conditions include education and comprehensive services for individuals with special needs, ongoing management of genetic conditions, access to medical devices, and referral to genetic support groups. As with the other recommended prevention activities, these services are available in Virginia but accessibility varies.

The CORN guidelines regarding research include a role for the state genetic coordinator and advisory committee in facilitating research awareness among professionals and the public as well as coordinating participation in genetic public health research projects. The Virginia system has generally not supported these activities, although genetic providers in the state may be involved with this on the local level. Educational activities at several levels, including general awareness, basic information, detailed instruction, and comprehensive training, are discussed in the CORN guidelines. While all are necessary for optimal genetic service provision, the recommendations call for public health genetic units to be involved primarily with the education of the public, teachers, non-genetic health care providers, and others through general awareness activities—such as flyers, news conferences, and media outlets—as well as basic information activities, including workshops and brochures. The Virginia Genetics Program has limited resources to undertake these activities; however, VDH has previously funded an annual genetics education conference and a November 2004 conference entitled *Genetic Horizons: Integrating Genetics into Public Health,* which was made possible through the Virginia CATPIP grant. The birth defects registry, VaCARES, is also in the process of developing educational brochures for consumers. Other groups, including the MOD, periodically coordinate educational activities regarding select genetic topics as well.

While Virginia appears to have a solid foundation of genetic services based on this comparison to CORN guidelines, unmet needs remain. Improvements in the coordination of services and program development are needed, while QA and further needs assessment should remain priorities for future initiatives. Optimally, planning to meet these needs would be included in a comprehensive state genetics plan. Comprehensive planning should include the development of measurable goals and objectives, with an expected date of completion where appropriate; an identifiable sequence of actions for achieving those objectives; identified resources necessary to meet the goals; estimates of the cost and benefits of the plan; a method for revision of the plan as objectives are reached or altered; and guidelines regarding plan priorities.

## Table 11-1. State Assessment Tool

### Comparing State Genetics Services to the CORN Guidelines for Clinical Genetic Services for the Public's Health

| I. ORGANIZATION & ADMINISTRATION | *In place* | *Planned* | *Undecided* |
|---|---|---|---|
| A. **Does the state health agency employ a full-time State Genetics Coordinator?** | ☑ | ☐ | ☐ |
| B. **Is there a State Genetics Advisory Board?** | ☑ | ☐ | ☐ |
| C. **Is there a State Plan for Provision of Genetic Services?** | ☐ | ☑ | ☐ |
| D. **Is the plan organized by public health core functions?** | ☐ | ☐ | ☑ |
| 1. **Assessment** - *Does the plan describe:* | | | |
| a. The state (size, geography, industry, etc.) | ☐ | ☐ | ☐ |
| b. Demographic parameters (age, race, socioeconomic status, etc.) | ☐ | ☐ | ☐ |
| c. Public health & genetics-related systems | ☐ | ☐ | ☐ |
| d. Data collection system | ☐ | ☐ | ☐ |
| e. System for evaluation of genetics services | ☐ | ☐ | ☐ |
| f. System for evaluation of educational activities | ☐ | ☐ | ☐ |
| g. Other: | ☐ | ☐ | ☐ |
| 2. **Policy Development** - *Does the plan describe:* | ☐ | ☐ | ☐ |
| a. Legislation in place related to genetics | ☐ | ☐ | ☐ |
| b. Mechanisms of funding/reimbursement for genetic services | ☐ | ☐ | ☐ |
| c. Legislation in place related to genetics | ☐ | ☐ | ☐ |
| 3. **Assurance** - *Does the plan include strategies to assure:* | ☐ | ☐ | ☐ |
| a. A network of genetic services | ☐ | ☐ | ☑ |
| b. A system of prevention services | ☐ | ☐ | ☑ |
| c. Genetics education activities | ☐ | ☐ | ☑ |
| d. Periodic review of genetic services | ☐ | ☐ | ☑ |
| e. A framework for existing quality assurance measures for clinical and laboratory services | ☐ | ☐ | ☑ |
| f. Other: | ☐ | ☐ | ☐ |
| 4. **Collaboration With Relevant Groups** | ☐ | ☐ | ☐ |
| a. Consumers | ☐ | ☑ | ☐ |
| b. Researchers | ☐ | ☐ | ☑ |
| c. Genetics Services Providers | ☐ | ☑ | ☐ |
| d. Teachers | ☐ | ☐ | ☑ |
| e. Other: | ☐ | ☐ | ☐ |
| E. **Linkages with Other State Programs** | ☐ | ☐ | ☐ |
| 1. Chronic Disease | ☐ | ☐ | ☑ |
| 2. Epidemiology | ☐ | ☐ | ☑ |
| 3. Other: | ☐ | ☐ | ☐ |

Note: Section I-D addresses the content of the plan for the state genetics program, not whether or not a specific program actually exists.

| **II. SERVICES** | | | | | |
|---|---|---|---|---|---|
| A. **Family-Based Services** | ***Readily available to ALL state residents*** | ***Readily available to MOST state residents*** | ***Readily available to SOME state residents*** | ***NOT readily available to state residents*** | ***Uncertain availability*** |
| 1. General Genetic Clinics | ☐ | ☑ | ☐ | ☐ | ☐ |
| 2. Metabolic Clinics | ☐ | ☐ | ☑ | ☐ | ☐ |
| 3. Single Disease Clinics | ☐ | ☐ | ☐ | ☐ | ☐ |
| a. Hemophilia | ☐ | ☐ | ☑ | ☐ | ☐ |
| b. Cystic Fibrosis | ☐ | ☐ | ☑ | ☐ | ☐ |
| c. Spina bifida | ☐ | ☐ | ☑ | ☐ | ☐ |
| d. Craniofacial | ☐ | ☐ | ☑ | ☐ | ☐ |
| e. Sickle cell anemia | ☐ | ☐ | ☑ | ☐ | ☐ |
| f. Muscular dystrophy | ☐ | ☐ | ☑ | ☐ | ☐ |
| g. Huntington disease | ☐ | ☐ | ☑ | ☐ | ☐ |
| h. Cancer | ☐ | ☐ | ☑ | ☐ | ☐ |
| 4. Prenatal Genetics Clinics | ☐ | ☑ | ☐ | ☐ | ☐ |

| B. **Population-Based Services** | ***Readily available to ALL state residents*** | ***Readily available to MOST state residents*** | ***Readily available to SOME state residents*** | ***NOT readily available to state residents*** | ***Uncertain availability*** |
|---|---|---|---|---|---|
| 1. Prenatal Screening | ☐ | ☐ | ☐ | ☐ | ☐ |
| a. Maternal serum | ☐ | ☑ | ☐ | ☐ | ☐ |
| b. Maternal age | ☐ | ☑ | ☐ | ☐ | ☐ |
| c. Family history | ☐ | ☑ | ☐ | ☐ | ☐ |
| d. Carrier screening for targeted populations | ☐ | ☑ | ☐ | ☐ | ☐ |
| 2. Newborn Screening | ☐ | ☐ | ☐ | ☐ | ☐ |
| a. Lab Services | ☑ | ☐ | ☐ | ☐ | ☐ |
| b. Follow Up | ☑ | ☐ | ☐ | ☐ | ☐ |
| 3. Birth defects monitoring | ☐ | ☐ | ☐ | ☐ | ☐ |
| a. Registry | ☑ | ☐ | ☐ | ☐ | ☐ |
| b. Follow Up | ☐ | ☐ | ☑ | ☐ | ☐ |
| 4. Teratogen monitoring | ☐ | ☐ | ☐ | ☐ | ☐ |
| a. Information service | ☐ | ☐ | ☐ | ☑ | ☐ |
| b. Outcome evaluation | ☐ | ☐ | ☐ | ☑ | ☐ |
| 5. Stillbirth evaluation | ☐ | ☐ | ☑ | ☐ | ☐ |
| 6. Childhood Screening | ☐ | ☑ | ☐ | ☐ | ☐ |
| 7. Adult Screening | ☐ | ☐ | ☐ | ☑ | ☐ |

| C. **Clinical Laboratory Services** | ***Available In-State*** | ***Coordinated with Out-of-State Lab*** | ***Not Available/ Not Coordinated*** |
|---|---|---|---|
| 1. Cytogenetics | ☑ | ☑ | ☑ |
| 2. Biochemical Genetics | ☑ | ☑ | ☑ |
| 3. Molecular Genetics | ☑ | ☑ | ☑ |

| D. **Modes of Service Delivery** | ***Check all that apply*** |
|---|---|
| 1. Genetics Unit of State Health Department | ☐ |
| 2. Large, comprehensive genetics center | ☑ |
| 3. Genetics unit of a comprehensive managed care facility | ☐ |
| 4. Resident genetics unit within a primary health care facility | ☐ |
| 5. Resident board certified genetic counselor and/or PhD medical geneticist with periodic visits by a board certified MD Medical Geneticist | ☐ |
| 6. Periodic visits by a board certified genetic counselor, medical geneticist, or other staff with local coordinators at outreach clinics | ☑ |
| 7. Genetics clinics in the private sector conducted by trained MD geneticists | ☑ |
| 8. Board certified genetic counselor and/or PhD medical geneticist within single disease/medical specialty setting | ☑ |
| 9. Other: Resident board certified genetic counselor | ☑ |

| E. **Genetics Professionals** | ***Readily available to ALL state residents*** | ***Readily available to MOST state residents*** | ***Readily available to SOME state residents*** | ***NOT readily available to state residents*** | ***Uncertain availability*** |
|---|---|---|---|---|---|
| 1. Clinical Geneticist | ☐ | ☑ | ☐ | ☐ | ☐ |
| 2. PhD Medical Geneticist | ☐ | ☐ | ☑ | ☐ | ☐ |
| 3. Genetic Counselor | ☐ | ☑ | ☐ | ☐ | ☐ |
| 4. Clinical Cytogeneticist | ☐ | ☐ | ☑ | ☐ | ☐ |
| 5. Clinical Biochemical Geneticist | ☐ | ☐ | ☑ | ☐ | ☐ |
| 6. Clinical Molecular Geneticist | ☐ | ☐ | ☑ | ☐ | ☐ |
| 7. Cytogenetic Technologist | ☐ | ☐ | ☑ | ☐ | ☐ |
| 8. Genetics Nurse | ☐ | ☐ | ☐ | ☑ | ☐ |
| 9. Advance Practice Nurse in Genetics | ☐ | ☐ | ☐ | ☑ | ☐ |
| 10. Perinatologist/Obstetrician | ☐ | ☑ | ☐ | ☐ | ☐ |
| 11. Other: | ☐ | ☐ | ☐ | ☐ | ☐ |

Note: Section II addresses the status of the programs and activities themselves, not just whether the issues are addressed in the state plan.

| **III. FUNDING OF SERVICES** | ***Check all that apply*** |
|---|---|
| A. Medicaid, Medicare reimbursement | ☑ |
| B. Third Party Carriers reimbursement | ☑ |
| C. Newborn Screening Surcharge | ☑ |
| D. State General Funds | ☑ |
| E. Federal Title V (MCH Block Grant) | ☑ |
| F. Other State and/or Federal Grants | ☑ |
| G. Specific Disease-Related Organizations | ☐ |
| H. Other: | ☐ |

| **IV. DOCUMENTATION OF NEEDS & SERVICES** | | | |
|---|---|---|---|
| A. **Data Sources** | ***Available and Utilized*** | ***Available and Not Utilized*** | ***Not Available*** |
| 1. State level clinical genetics database | ☐ | ☐ | ☑ |
| 2. Newborn screening database | ☑ | ☐ | ☐ |
| 3. Vital statistics: birth, fetal death, death certificates | ☑ | ☐ | ☐ |
| 4. Statewide hospital discharge data | ☑ | ☐ | ☐ |
| 5. Medicaid/Medicare eligibility, claims, provider datasets | ☐ | ☑ | ☐ |
| 6. Local/Statewide/Regional cytogenetics registry | ☐ | ☐ | ☑ |
| 7. Local/Statewide/Regional birth defects registry | ☑ | ☐ | ☐ |
| 8. Local/Statewide/Regional population based cancer/tumor registry | ☑ | ☐ | ☐ |
| 9. Directory of genetic service providers and referral sources | ☑ | ☐ | ☐ |
| 10. Cytogenetics laboratory databases collected by ACT | ☐ | ☑ | ☐ |
| 11. Federal census data | ☑ | ☐ | ☐ |
| 12. Special surveys and projects: | ☐ | ☐ | ☐ |
| a. Pregnancy Risk Assessment Monitoring System (PRAMS) | ☑ | ☐ | ☐ |
| b. National Maternal & Infant Health Survey | ☐ | ☑ | ☐ |
| c. Behavioral Risk Factor Surveillance System (BRFSS) | ☑ | ☐ | ☐ |
| d. National Survey of Family Growth | ☐ | ☐ | ☑ |
| 13. Other: | ☐ | ☐ | ☐ |

| B. **Data Linkages** | ***Linkage in Place*** | ***Planned Linkage*** | ***Linkage Under Consideration*** | ***Linkage Not Under Consideration*** |
|---|---|---|---|---|
| 1. Birth & death for all deaths up to six years of age | ☐ | ☑ | ☐ | ☐ |
| 2. Birth defects & tumor registry for all pediatric cancer cases | ☐ | ☐ | ☑ | ☐ |
| 3. Birth defects registry records with vital statistics | ☐ | ☑ | ☐ | ☐ |
| 4. Inpatient hospital discharge records with birth certificates | ☐ | ☑ | ☐ | ☐ |
| 5. Newborn screening records with birth certificates | ☐ | ☐ | ☑ | ☐ |
| 6. MSAFP/AFAFP/Triple Screen with vital statistics | ☐ | ☐ | ☐ | ☑ |
| 7. Statewide clinical genetics services database and birth/fetal death certificates (in form of numerator/denominator ratio) | ☐ | ☐ | ☐ | ☑ |
| 8. System for direct referral from clinical genetics to early intervention services for infants < 3 years of age; CSHCN; Supplemental Social Insurance (SSI); etc. | ☐ | ☐ | ☑ | ☐ |
| 9. Other: | ☐ | ☐ | ☐ | ☐ |

**Table 11-2. CORN's Recommended State Genetics Unit Responsibilities (2)**

- Facilitate communication within the genetics community in the state.
- Maintain linkages between the genetics community, consumers, and all relevant components of the state department of health.
- Maintain familiarity with all aspects of clinical and laboratory genetic services, including:
  - prevention
  - dissemination of information/training and education programs
  - needs and resources
  - mechanisms of reimbursement
- Understand the distribution of genetic services in the state and promote
- Identify needs for additional genetic services.
- Understand existing data collection programs and address additional needs.
- Monitor state legislation and regulatory efforts relevant to genetics.
- Maintain familiarity with recognized professional standards for clinical and laboratory personnel, facilities, and genetic services.
- Monitor all contracts related to state-funded genetic services.
- Collaborate closely with the state genetics advisory council.

## 12. INSIGHTS FROM OTHER STATES

Many other states have conducted needs assessments and developed state genetic plans, including nineteen that were reviewed over the course of this project. Most of these documents are available through links on the NNSGRC Web site http://genes-r-us.uthscsa.edu/resources/genetics/geneticsplan.htm. While genetic services in Virginia are influenced by some factors unique to the Commonwealth, the experiences of other states can serve as useful resources for Virginia's leaders as they move forward with efforts to improve the genetic systems of care for all Virginians. Available data comparing select Virginia systems with other states is reviewed here, along with brief summaries of inventive programs developed by other states in response to identified needs, many of which mirror gaps in the Virginia system.

Although there are limited data comparing state genetic systems, two studies have assessed birth defect registries and population-based genetic databases throughout the U.S. In 2002, Trust for America's Health (TFAH), a national non-profit organization, conducted a nationwide assessment of birth defect registries using a number of criteria, including the ability to carry out tracking, data use, prevention and research capacity, data sharing capacity and resources devoted to the task. Overall, many states and territories were found to have inadequate, or in some cases, non-existent programs. In particular, the majority of programs do not facilitate the identification of links between birth defects and environmental exposures, considered a primary purpose of such registries. Virginia's birth defect surveillance program, VaCARES, was in the top half of states, with 30 programs receiving lower rankings. Although many aspects of VaCARES exceeded minimum standards, gaps identified included timeliness of data publication, non-inclusion of fetal deaths, lack of complete population coverage, and poor funding (1). An ongoing grant-funded project, Virginia CATPIP, is aimed at improving VaCARES, progress that was noted by TFAH in its most recent review (2). A study published in 2000 revealed that population-based databases across the country that are relevant to public health genetics—including birth defect surveillance, clinical genetic services, newborn screening, and CSHCN databases—are limited and are poorly integrated into program activities where they do exist, hindering their usefulness for planning, monitoring, or evaluating genetic programs in their respective states (3). Significant variations in public health genetics programs exist in the U.S., varying from the comprehensive, well integrated *Genetic Disease Branch* in California with a staff of 129 and an annual budget of $60 million in 1997, to programs limited to newborn screening, to virtually non-existent programs (4). Perhaps more applicable as models for Virginia are systems in states with similar population levels, including Iowa, with a well organized public health genetics system that has been in place for a number of years, and Missouri, with a staff of nine in the state genetics unit (5, 6).

A review of other states' needs assessment reports and published genetic plans revealed significant variations in their methods, utility, and thoroughness, with some very limited in scope, while others provided comprehensive evaluations of services and programs. Most of the comprehensive plans were based on studies that included primary data collected during the needs assessment process, as is underway in Virginia. While Virginia's assessment includes feedback from genetic providers, PCPs, and consumers in the state, some other states collected data from a broader range of stakeholders, as in Oregon, where 22 different assessment activities included public health faculty, medical billing specialists, educators, college students, and many other stakeholder groups (7, 8). The needs assessment in Texas included a legal review of all current and proposed legislation and rulings that affect genetic activities in the state by an attorney specializing in health law and policy, while Washington's assessment included a random telephone survey of the public (9, 10). Overall, a broad-based evaluation of genetic service needs appears to require feedback from genetic providers, non-genetic providers, and consumers at minimum. A variety of standards were used to assess genetic systems in different states. Although many states used the CORN

guidelines as their primary evaluation tool, others used the *Ten Essential Public Health Services* (Table 12-1), developed by major public health organizations in 1994 as an enhancement of the core public health functions of assessment, policy development, and assurance (11). Some states used an elaboration of these criteria, developed by ASTHO specifically for public health genetics, to assess their programs, while Washington used the *Healthy People 2010* objectives as a model when developing their current genetic plan (12, 13). Another hallmark of the truly comprehensive, useful state genetic plans appears to be the involvement of a broad range of stakeholders in drafting the document, as exemplified by the group that developed the Oklahoma plan, the Oklahoma Genetics Advisory Council, which included genetic and other health care providers, state public health leaders, consumers, state health professional organization leaders, health administrators, ethicists, clergy, legislators, and insurance representatives, among others (7). The presentations of their genetic plans vary significantly among states as well. Some states, such as Ohio and Oklahoma, developed documents that are useful for consumer education and program marketing, while others are presented as expert planning resources for ongoing program development (7, 14). The Indiana state plan, for example, delineates clearly defined goals that are further elaborated in objectives to be accomplished through concrete action steps with target completion dates (15). These tools and models provide potential foundations on which to build a plan for Virginia.

**Table 12-1.Ten Essential Public Health Services**

| |
|---|
| 1. Monitor health status to identify community health problems. |
| 2. Diagnose and investigate health problems and health hazards in the community. |
| 3. Inform, educate, and empower people about health issues. |
| 4. Mobilize community partnerships to identify and solve health problems. |
| 5. Develop policies and plans that support individual and community health efforts. |
| 6. Enforce laws and regulations that protect health and ensure safety. |
| 7. Link people to needed health services and assure the provision of health care when otherwise unavailable. |
| 8. Assure a competent public health and personal health workforce. |
| 9. Evaluate effectiveness, accessibility, and quality of personal and population-based health services. |
| 10. Research for new insights and innovative solutions to health problems. |

Several areas were identified as significant needs by multiple states, and are likely to be priority areas for Virginia as well. Identified gaps most commonly involved data integration, access to services, education, and privacy/confidentiality issues. As in Virginia, manpower deficits were identified by virtually all states as significant barriers impeding access to genetic services. Particularly with the expansion in understanding of the genetic basis of common disease as well as the growing impact of genetics throughout all areas of medicine, states should develop strategies to minimize genetic provider shortages in the future (15, 16). Some states also identified a need for the development or expansion of teratogen information services, newborn screening programs, and transitional and long-term care services for adolescents and adults in CSHCN programs, areas that Virginia may want to examine further (17).

Several states have developed innovative programs to meet identified needs, a few of which are highlighted here. In Colorado, the Community Notification and Referral Program, developed as part of the birth defect surveillance system, links infants referred to the birth defect registry with local public health and early childhood agencies which inform the families of services and resources in the community, including CSHCN programs (18). The TIPS (Tracking Infant Progress) program in Nebraska provides central identification and tracking of infants admitted to NICUs statewide, facilitating early identification and referral of many children with special health care needs (17). The North Carolina

Genetic Health Care Branch within the state's Division of Women's and Children's Health supports six regional public health genetic counselors and nine sickle cell educators to facilitate communication with the comprehensive genetic centers and provide coordinated patient services (4).

The one gap identified as a major priority by all the state assessments is the need for increased public and professional education in genetics. Washington and Oklahoma have developed the most broad-based, comprehensive education programs in response to this identified need, both of which would be very helpful resources for the future development of a statewide education program in Virginia. In Washington, a Genetics Education Steering Committee was created, followed by the development of the *Genetic Education Project (GEP)* focused on educating PCPs and others about medical genetic services and interventions using a train-the-trainer model and a curriculum developed jointly with the MOD, *Genetics and Your Practice* (4, 19). This education plan includes recommendations applicable to all groups, as well as a review of information needs, current programs, challenges, and suggestions for future programs for a variety of specific target audiences, including:

- Adoption Workers
- Affected Families
- Allied Health Care Professionals
- Clergy
- Clinical Genetics Professionals
- Genealogists
- General Public
- Insurance Providers And Purchasers
- Laboratorians
- Law Makers
- Legal Professionals
- Librarians
- Medical and Professional Association Leaders
- News Media
- Physicians
- Public Health Professionals
- Researchers
- Support Groups
- Teachers

Oklahoma, which has been awarded a four year, $1.2 million grant from the MCHB of HRSA to implement their state genetics plan, has identified education as a primary goal of the plan. A genetic counselor acts as the State Genetics Education Coordinator for the project, which involves establishing a genetics education resource center, conducting an education campaign, maximizing genetics education opportunities, and developing a statewide program of preconception and prenatal health education. The campaign targeting health care providers includes presentations to a variety of specialists, medical publications, and development of a Web site that includes a genetics referral guide. Genetic presentations and targeted handouts have been developed for high school students and biology teachers as well. Policymakers have been educated through meetings with the Secretary of Health and the distribution of legislative information relevant to genetics. The Web site being developed as part of the project includes areas aimed at education of the public, which has also been facilitated through workshops, support group presentations, and public events related to genetics. In Virginia, the Web site maintained by the genetics program within VDH includes similar public educational materials, as well as links to other information sources. The Oklahoma project has included other target groups through a variety of initiatives in this broad-based, comprehensive education campaign (20).

NYMAC (New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services) was established in September 2004 as one of seven regional collaboratives in the country funded by the Genetic Services Branch in the Health Resources and Services Administration (HRSA)'s Maternal and Child Health Bureau. The charge of this group is to develop a regional approach to address the unequal distribution of genetic resources in the New-York-Mid-Atlantic region, which includes Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia and West Virginia. This group has taken on some of the CORN functions on a regional level with working groups in distribution of services, educational issues, laboratory and newborn screening issues, consumer concerns and other areas of both state and regional importance. Members of the genetics community and consumers from Virginia are involved in these activities and will foster sharing of information.

While significant resources will be necessary, programs and efforts such as those described here can facilitate improvement in the genetic health of Virginians, as well. Along with the planned collection of feedback from other stakeholders in Virginia, this needs assessment data provides a rich source with which to design a useful state genetic plan. However, other resources will be necessary for the development of a truly comprehensive plan. These include the participation of a diverse working group, as in the Oklahoma model, as well as a dedication to the delineation of well defined goals and action steps, as exemplified by the Indiana plan. Most of the states highlighted here conducted their needs assessment activities and developed genetic plans over the course of two to three years, often with grant funding from HRSA. Virginia’s leaders must be resourceful in obtaining funding for the development and implementation of a similarly useful plan for the Commonwealth that meets the genetic needs of her citizens.

# 13. THE COMMONWEALTH POLL: VIRGINIANS' AWARENESS OF AND ATTITUDES TOWARD GENETIC SERVICES

## Overview

In an effort to collect the thoughts and opinions of the citizens of the Commonwealth of Virginia, a contract was established with the Virginia Commonwealth University Center for Public Policy to conduct a Commonwealth Poll. The Commonwealth Poll is an omnibus public opinion survey of Virginia residents. Each survey covers a variety of topics. The survey is conducted by telephone with a randomly-selected sample of adult Virginians. The questions for this poll were designed by the research team with assistance from the VCU Center for Public Policy to identify the general public's knowledge of genetics; their usage of genetic services; the information sources they used to find out about genetic services; issues regarding the location of genetic centers; and their perceptions of the importance of having genetic services available to Virginians. The completed poll questions are included as Appendix E.

Interviewing was conducted from June 17 to July 8, 2005, following approval from the VCU Institutional Review Board (IRB), by telephone from the facilities of the Survey and Evaluation Research Laboratory at Virginia Commonwealth University in Richmond. The interviewing was conducted by a staff of professionally trained, paid interviewers using computer-assisted telephone interviewing software.

The sample of telephone numbers was prepared by Genesys Sampling Systems of Ft. Washington, Pennsylvania, and was designed so that all residential telephones, including new and unlisted numbers, had a known chance of inclusion. The cooperation rate for the survey was 35% percent. Using the Council of American Survey Research Organization (CASRO) response rate calculations, interviews were obtained with respondents in 29% percent of the known or assumed residential households in the sample. The final survey sample consisted of 801 respondents. The respondents represented a broad cross-section of Virginians, based on information gathered about geographic region, age, sex, race, education, and income.

The data were weighted to adjust for unequal probabilities of selection due to multiple telephone lines and multiple adults living in the household. In addition, the data were weighted on sex, race, age, and region of residence to reflect the demographic composition of the Virginia adult population. Percentages reported in the text and tables are weighted, while the number of cases shown in the tables for various subgroups is the actual number of respondents.

Questions answered by the full sample of adults are subject to a sampling error of plus or minus approximately 3.5 percentage points at the 95 percent level of confidence. This means that in 95 out of 100 samples like the one used here, the results obtained should be no more than 3.5 percentage points above or below the figure that would be obtained by interviewing all adult Virginians with telephones. Where the answers of subgroups are reported, the sampling error would be higher. Because of nonresponse (refusals to participate, etc.), standard calculations of sampling error are apt to understate the actual extent to which survey results are at variance with the true population values. Surveys are also subject to errors from sources other than sampling. While every effort is made to identify such errors, they are often difficult or impossible to measure. Readers making use of the results are urged to be mindful of the limitations inherent in survey research.

## Knowledge of Genetics

Respondents were asked to rate their personal knowledge of genetics on a scale of on a scale of 1 to 5, with 1 being "I know almost nothing at all," and 5 being "I know almost everything I need to know." Responses were spread across the five knowledge categories; 32 percent of the respondents rated their knowledge level with a 4 or 5, 41 percent rated their knowledge level with a 3, and 26 percent rated their knowledge level with a 1 or 2. The average knowledge score was 3.10 out of a maximum score of 5.00

### Table 13-1. Personal Genetic Knowledge

**How would you rate your own personal knowledge of genetics on a scale of 1 to 5 where 1 means "I know almost nothing at all" and 5 means "I know everything I need to know"?**

| | | 1 | 2 | 3 | 4 | 5 | Do not know | No answer | Number of cases |
|---|---|---|---|---|---|---|---|---|---|
| Region | Northwest | 9% | 19 | 42% | 15% | 15% | | | 135 |
| | Northern VA | 6% | 19 | 45% | 17% | 12% | | 1% | 179 |
| | West | 16% | 15 | 30% | 22% | 16% | 1% | | 156 |
| | South Central | 7% | 14 | 43% | 26% | 8% | 1% | 2% | 163 |
| | Tidewater | 5% | 19 | 45% | 20% | 10% | 0% | | 168 |
| Age | 18-29 | 8% | 13 | 48% | 23% | 8% | | | 97 |
| | 30-44 | 7% | 22 | 41% | 17% | 13% | | | 203 |
| | 45-64 | 8% | 16 | 40% | 22% | 12% | 1% | 0% | 349 |
| | 65 and older | 10% | 19 | 36% | 18% | 14% | 1% | 2% | 114 |
| Sex | Male | 9% | 18 | 40% | 22% | 10% | 0% | 0% | 341 |
| | Female | 7% | 17 | 42% | 18% | 14% | 1% | 1% | 460 |
| Race | White | 8% | 20 | 41% | 19% | 11% | 0% | 1% | 628 |
| | Non-white | 7% | 11 | 43% | 26% | 12% | 1% | | 138 |
| Hispanic or Spanish origin? | Yes | 11% | 24 | 33% | 15% | 17% | | | 24 |
| | No | 8% | 17 | 42% | 21% | 11% | 0% | 1% | 765 |
| Education | No high school diploma | 16% | 20 | 33% | 17% | 10% | 2% | 2% | 49 |
| | High school diploma | 19% | 20 | 32% | 14% | 13% | 1% | 1% | 151 |
| | Some college | 6% | 20 | 43% | 17% | 13% | | 1% | 218 |
| | College graduate or more | 3% | 15 | 46% | 25% | 11% | 0% | | 372 |
| Family income | Under $20,000 | 18% | 11 | 36% | 19% | 16% | | | 55 |
| | $20,000-$34,999 | 12% | 23 | 33% | 16% | 13% | 2% | 1% | 88 |
| | $35,000-49,999 | 9% | 14 | 47% | 15% | 14% | | | 104 |
| | $50,000-69,999 | 7% | 26 | 35% | 20% | 11% | 1% | 1% | 127 |
| | $70,000 and above | 5% | 15 | 47% | 22% | 11% | | | 299 |

## Use of Genetic Services

Respondents provided information about their use of genetic services over the last five years. Ten percent

of the respondents reported that they had used any genetic services in the last five years. Those who used genetic services were asked which services they had used, and they could list as many services as they could remember using. The most frequently cited services used were: the evaluation or diagnosis of birth defects or genetic diseases in children or adults (20 percent of all mentions); screening of a pregnancy for genetic diseases or birth defects (19 percent of all mentions); and genetic counseling (17 percent of all mentions). Among the respondents who had not used any genetic services in the last five years, over three-fourths (78 percent) said the reason was that they had no need for the services or that they were not interested in receiving the services. Of the remaining reasons given, the most frequent one—given by only 4 percent of the respondents—was that they did not know that services were available.

## Table 13-2. Use of Genetic Services

**Within the last 5 years, have you received any genetic services?**

| | | Yes | No | Do not know | No answer | Number of cases |
|---|---|---|---|---|---|---|
| Region | Northwest | 7% | 93% | | | 135 |
| | Northern VA | 16% | 84% | | 0% | 179 |
| | West | 7% | 93% | | | 156 |
| | South Central | 7% | 93% | | | 163 |
| | Tidewater | 11% | 88% | 1% | | 168 |
| Age | 18-29 | 5% | 95% | | | 97 |
| | 30-44 | 15% | 84% | 1% | | 203 |
| | 45-64 | 10% | 90% | | | 349 |
| | 65 and older | 7% | 93% | | | 114 |
| Sex | Male | 6% | 94% | 1% | 0% | 341 |
| | Female | 15% | 85% | | | 460 |
| Race | White | 11% | 89% | | 0% | 628 |
| | Non-white | 9% | 89% | 1% | | 138 |
| Hispanic or Spanish origin? | Yes | 18% | 82% | | | 24 |
| | No | 10% | 90% | | 0% | 765 |
| Education | No high school diploma | 8% | 92% | | | 49 |
| | High school diploma | 7% | 93% | | | 151 |
| | Some college | 9% | 90% | 1% | | 218 |
| | College graduate or more | 13% | 87% | | 0% | 372 |
| Family income | Under $20,000 | 9% | 91% | | | 55 |
| | $20,000-$34,999 | 2% | 98% | | | 88 |
| | $35,000-49,999 | 7% | 90% | 2% | | 104 |
| | $50,000-69,999 | 10% | 90% | | | 127 |
| | $70,000 and above | 15% | 85% | | | 299 |

**Table 13-3. Types of Services Used**

**Within the last five years, what kinds of genetic services have you received? (respondents could name multiple services)**

| | Number of Responses | % of Total Responses |
|---|---|---|
| Genetic counseling | 17 | 17% |
| Eval:diagnosis genetic disease/birth defect in child/adult | 20 | 20% |
| Treatment of genetic disease/birth defect in child/adult | 5 | 5% |
| Screening of a pregnancy for genetic disease/birth defect | 19 | 19% |
| Genetic testing in either a child or an adult | 11 | 11% |
| Newborn screening | 10 | 10% |
| Other | 17 | 17% |
| Do not know | 0 | 0% |
| No answer | 1 | 1% |
| Total | 100 | 100% |

## Information Sources Used

Less than one-third (28 percent) of all respondents reported that they had ever needed genetic information or had questions about genetics. Respondents who had needed genetic information were asked to list the sources of information they used to get their questions answered, and they could list multiple sources. The most frequently mentioned sources were: the Internet (23 percent of all mentions); the family physician/primary care physician (21 percent of all mentions); reference books (11%) of all mentions); and family (11% of all mentions).

All respondents were asked where they might go in the future to get answers, if they had questions about genetics. Again, the two most frequently mentioned sources were: the Internet (33 percent of all mentions); and the family physician/primary care physician (27 percent of all mentions).

## Privacy of Genetic Information

A majority of respondents (53 percent) indicated that they were “very concerned” or “somewhat concerned” about their ability to keep their genetic information private, while 45 percent were “not too concerned” or “not at all concerned.”

## Table 13-4. Genetic Privacy Concerns

**In general, how concerned are you about your ability to keep your genetic information private?**

| | | Very concerned | Somewhat concerned | Not too concerned | Not at all concerned | Do not know | No answer | Number of cases |
|---|---|---|---|---|---|---|---|---|
| Region | Northwest | 16% | 33% | 28% | 20% | 1% | 2% | 135 |
| | Northern VA | 23% | 27% | 34% | 15% | 1% | | 179 |
| | West | 34% | 28% | 21% | 14% | 2% | | 156 |
| | South Central | 28% | 24% | 28% | 17% | 3% | | 163 |
| | Tidewater | 24% | 28% | 31% | 15% | 2% | | 168 |
| Age | 18-29 | 21% | 35% | 30% | 13% | 2% | | 97 |
| | 30-44 | 29% | 31% | 26% | 13% | 1% | | 203 |
| | 45-64 | 24% | 25% | 32% | 16% | 2% | | 349 |
| | 65 and older | 26% | 14% | 35% | 23% | 2% | | 114 |
| Sex | Male | 25% | 27% | 29% | 18% | 2% | | 341 |
| | Female | 26% | 28% | 30% | 14% | 2% | | 460 |
| Respondent | White | 22% | 28% | 32% | 16% | 2% | | 628 |
| | Non-white | 34% | 27% | 21% | 15% | 1% | | 138 |
| Hispanic or Spanish origin? | Yes | 40% | 9% | 27% | 24% | | | 24 |
| | No | 25% | 28% | 29% | 15% | 2% | | 765 |
| Education | No high school diploma | 26% | 14% | 29% | 24% | 5% | 1% | 49 |
| | High school diploma | 27% | 24% | 27% | 19% | 2% | 1% | 151 |
| | Some college | 30% | 28% | 25% | 15% | 2% | | 218 |
| | College graduate or more | 22% | 30% | 33% | 14% | 1% | | 372 |
| Family income | Under $20,000 | 30% | 15% | 29% | 22% | 3% | 2% | 55 |
| | $20,000-$34,999 | 33% | 25% | 22% | 19% | 1% | | 88 |
| | $35,000-49,999 | 26% | 25% | 31% | 16% | 2% | 1% | 104 |
| | $50,000-69,999 | 23% | 27% | 35% | 14% | 1% | | 127 |
| | $70,000 and above | 23% | 28% | 31% | 16% | 1% | | 299 |

## Location of Genetic Centers

Only about 1 in 10 respondents (11 percent) said they knew the location of the genetic center nearest to their home. When all respondents were asked about the maximum number of miles they would be willing to travel in order to get services from a genetic center, 12 percent said 10 miles or less; 19 percent said 11 to 30 miles; 16 percent said 31 to 50 miles; and 33 percent said more than 50 miles. In fact, 22 percent of the respondents said they would be willing to travel more than 100 miles to get services from a genetic center.

## Importance of Genetic Services

Almost 9 in 10 respondents (86 percent) said that it was either “very important” or “somewhat important”

to have genetic services available to the people of Virginia. Only 11 percent said it was "not too important" or "not at all important."

**Table 13-5. Importance of Services**

**In your opinion, how important is it to have genetic services available to the citizens of Virginia?**

| | | Very | Somewhat | Not too | Not at all | Do not know | No answer | Number of cases |
|---|---|---|---|---|---|---|---|---|
| Region | Northwest | 40% | 42% | 7% | 4% | 4% | 3% | 135 |
| | Northern VA | 36% | 45% | 12% | 4% | 3% | 1% | 179 |
| | West | 42% | 47% | 5% | 1% | 4% | 1% | 156 |
| | South Central | 42% | 47% | 6% | 4% | 1% | 0% | 163 |
| | Tidewater | 36% | 49% | 9% | 2% | 3% | 1% | 168 |
| Age | 18-29 | 31% | 56% | 6% | 3% | 3% | 0% | 97 |
| | 30-44 | 41% | 48% | 6% | 3% | 1% | 1% | 203 |
| | 45-64 | 38% | 45% | 10% | 1% | 4% | 1% | 349 |
| | 65 and older | 46% | 35% | 8% | 4% | 4% | 2% | 114 |
| Sex | Male | 29% | 51% | 11% | 4% | 3% | 1% | 341 |
| | Female | 47% | 42% | 6% | 1% | 2% | 1% | 460 |
| Race | White | 37% | 46% | 9% | 3% | 3% | 1% | 628 |
| | Non-white | 45% | 50% | 1% | 2% | 1% | 1% | 138 |
| Hispanic or Spanish origin? | Yes | 54% | 32% | 3% | | 4% | 7% | 24 |
| | No | 38% | 47% | 8% | 3% | 3% | 1% | 765 |
| Education | No high school diploma | 46% | 42% | 5% | 1% | 5% | 1% | 49 |
| | High school diploma | 44% | 42% | 5% | 3% | 4% | 2% | 151 |
| | Some college | 36% | 51% | 6% | 2% | 3% | 2% | 218 |
| | College graduate or more | 37% | 47% | 11% | 3% | 2% | 0% | 372 |
| Family income | Under $20,000 | 53% | 34% | 6% | 2% | 5% | | 55 |
| | $20,000-$34,999 | 46% | 47% | 2% | 2% | 2% | 1% | 88 |
| | $35,000-49,999 | 39% | 47% | 10% | 2% | 1% | 1% | 104 |
| | $50,000-69,999 | 36% | 53% | 5% | 3% | 2% | 1% | 127 |
| | $70,000 and above | 37% | 46% | 11% | 3% | 2% | | 299 |

## Suggestions for Genetic Services

Ten percent of the respondents had suggestions for genetic services they would like to see in Virginia that are currently not offered. Among those suggestions were: more information and/or publicity about genetics (29 percent of all responses); additional genetic centers (17 percent of all responses); and more or better facilities (9 percent of all responses).

## Impact of These Findings

Among our respondents, 67% reported that their understanding of genetics was average or below what they felt was important to know. This is consistent with the findings in other state and international polls (1-4). This self-reported knowledge is based on the individual's perception of their level of understanding

and is not a measure of actual genetic literacy. A recent study has shown that while scientific literacy is on the rise, only 20-25% of Americans are "scientifically savvy and alert." Within this same study, less than a third could identify that DNA was a key to heredity (5).

Without a solid base of genetic understanding, including the limitations and risks associated with some genetic tests, individuals cannot truly make informed decisions about using genetic services. As genetics continues to evolve and more is recognized about the genetic basis of even common diseases, the number of families and individuals who may be impacted rises dramatically. Increased public education is therefore needed. In an open-ended question, 29% of those polled identified education as a way to improve the services offered in the Commonwealth of Virginia. This need for education has been consistently identified by each group involved within this needs assessment. It will be important in developing educational programs to remember to provide relevant facts that the public can translate into personally meaningful information.

Ten percent of our respondents did report that they had used some form of genetic service within the last five years. Interestingly, only 10 (1.2%) reported having used newborn screening. Though we do not know how many of those polled have children born within the last 5 years, 37.5% of the sample was under the age of 45, and therefore presumably of childbearing age. Within the Commonwealth of Virginia, newborn screening is a mandated test and the uptake is high. Given the low number of newborn screens reported, it is possible that many of those polled failed to recognize newborn screening as a form of genetic testing.

Of those who had not received genetic services within the last five years, 75% stated it was because services were not needed. While this is certainly a plausible explanation for many individuals, in numerous cases services may be beneficial or needed, but the individual fails to recognize the need. This reiterates an increasing need for additional education of the public. An understanding of genetic services precedes a perceived or realized need for these services.

Overall, 86% of respondents felt genetics services are important to the citizens of Virginia, though 53% do have remaining concerns about the privacy of their genetic information. Genetic privacy concerns have been found consistently in studies across the nation and these privacy issues will need to be addressed in educational programs that are developed (1, 3, 4, and 6). In addition to the need for education, the public did also identify a need for additional and better genetic centers to provide the citizens with geographically accessible services. These needs again echo the needs identified by the other groups included within this needs assessment.

# 14. A SURVEY OF KEY STAKEHOLDERS

## Overview

In an effort to continue the assessment of the current and emerging practices and deficiencies within genetic health services and education, a survey was developed to collect the opinions and feedback of other key stakeholders. The involvement of many individuals and groups in this thoughtful needs assessment will help to energize the collective whole to address unmet needs in genetics for Virginia's citizens. The key stakeholders polled in this final survey included representatives of insurance companies, specialty doctors (i.e. oncologists, surgeons, cardiologists), allied healthcare professionals (i.e. nurses, nurse practitioners), major employers in Virginia, representatives of regional hospitals, medical schools, and organizations, government representatives, representatives from the education system, and the media.

Questions included in the stakeholder survey were developed with input from the VaGAC and included questions addressing their knowledge of genetics, their opinion on the average Virginian's knowledge of genetics, and the current status of genetic services. The complete survey is included in Appendix F. Following approval by the VCU Institutional Review Board (IRB), the survey was distributed by postal mail to 150 stakeholders within the identified groups. Phone numbers were provided to recipients in case they wished to address any questions or topics in greater detail. A reminder copy of the survey was sent three weeks after the original to maximize response rates. Ten surveys were returned due to an old or incorrect mailing address and a new address was unavailable. Thirty-seven surveys were received for a response rate of 29%. Not all respondents answered every question. A comparison of respondents and non-respondents is unavailable.

## Knowledge of Genetics

Respondents were asked to rate their personal knowledge of genetics. Twenty-three percent felt their own knowledge was "poor" or "very poor," while 46% rated their knowledge as "good" or "excellent". Thirty percent felt they had a "neutral" knowledge level.

In comparison, when asked to rate the genetic knowledge level of the average Virginian on the same scale, 78% listed "poor" or "very poor" and 0% listed "good" or "excellent". Twenty percent identified the average Virginian's knowledge as "neutral." Seventy-eight percent felt that the average Virginian even lacked the knowledge to know where to go to have their genetic-based questions answered.

**Table 14-1. Knowledge of Genetics**

| | **Personal Knowledge** | | **Average Virginian's Knowledge** | |
|---|---|---|---|---|
| | Number of Responses | % of Total Responses | Number of Responses | % of Total Responses |
| Very Poor | 1 | 3 | 10 | 25 |
| Poor | 8 | 20 | 21 | 53 |
| Neutral | 12 | 30 | 8 | 20 |
| Good | 17 | 43 | 0 | 0 |
| Excellent | 1 | 3 | 0 | 0 |

* Percentages may not equal 100% due to rounding and incomplete responses on some surveys.

Also included in the survey was a question to assess our respondent's awareness of folic acid's role in birth defects prevention. The prevention of birth defects is certainly one important goal of genetic services. Education efforts related to the benefits of folic acid have been ongoing for years and have been a major focus of the March of Dimes. The stakeholders were specifically asked in order to reduce the risk of neural tube defects when a woman should be taking 0.4 mg of folic acid. Overall, 58% answered correctly, while 43% answered incorrectly or failed to answer.

**Table 14-2. Folic Acid Awareness**

**In order to reduce the risk of neural tube defects a woman should be taking 0.4mg of folic acid daily if she is:**

| | Number of Responses | % of Total Responses |
|---|---|---|
| Planning a Pregnancy | 10 | 25 |
| Capable of Becoming Pregnant | 2 | 5 |
| May Become Pregnant in the Future | 1 | 3 |
| All of the Above[1] | 23 | 58 |
| No Answer Provided | 4 | 10 |

* Percentages may not equal 100% due to rounding and incomplete responses on some surveys.
[1] Correct answer

## The Importance and Need for Genetic Services

Overwhelmingly, respondents felt that having genetic services available to the citizens of Virginia was important. Eighty-six percent felt the availability of services was "very important" or "somewhat important." Ten percent were "neutral" on this issue. Zero respondents felt that services were "somewhat unimportant" or "very unimportant."

When questioned about the current status of genetic services in Virginia, there were a surprisingly high number of key stakeholders who felt uninformed on the issues. Thirty percent felt there are problems with the current accessibility or availability of genetic services. Eight percent felt there are no issues with accessibility, and 63% did not know. Similarly, 0% felt that there are currently enough genetic providers, 18% responded that the number of providers is lacking, and 83% did not know. In terms of the geographic distribution of the practicing providers, 0% felt there was adequate distribution, 15% felt the distribution was inadequate, and 85% did not know.

Equally high numbers were uninformed on the insurance coverage and privacy of genetic services. Five percent replied that genetic services are adequately covered by most health insurance plans, 35% felt coverage was lacking, and 60% did not know. Similarly, while 30% of those surveyed felt that the level of protection current genetic discrimination and privacy laws provides for Virginians was adequate, with only 8% being unsatisfied with current protections, 63% did not know.

**Table 14-3. The Current Status of Genetic Services**

| | Accessibility Problems | | Sufficient Number of Providers | | Sufficient Geographic Distribution of Providers | | Adequate Insurance Coverage | |
|---|---|---|---|---|---|---|---|---|
| | Number of Responses | % of Total Responses | Number of Responses | % of Total Responses | Number of Responses | % of Total Responses | Number of Responses | % of Total Responses |
| Yes | 12 | 30 | 0 | 0 | 0 | 0 | 2 | 5 |
| No | 3 | 8 | 7 | 18 | 6 | 15 | 14 | 35 |
| Don't Know | 25 | 63 | 33 | 83 | 34 | 85 | 24 | 60 |

* Percentages may not equal 100% due to rounding and incomplete responses on some surveys.

## Future Needs for Genetic Services in Virginia

These key stakeholders were asked to identify the top three genetic needs for Virginians. The top needs identified were a need for more genetic education (63%), more application of genetic research to clinical problems (45%), a need for more educational materials for families (35%) and a need for more public discussion of ethical, social, and legal issues surrounding genetics (35%). Also frequently mentioned were more genetic research (23%), improved coordination of services (20%), telemedicine capabilities for genetic consultations (18%), a need for more providers (15%), and increased newborn screening tests (15%).

**Table 14-4. Genetic Needs for Virginians**

| | Number of Responses | % of Total Responses |
|---|---|---|
| Genetic Education | 25 | 63 |
| Application of Research to Clinical Problems | 18 | 45 |
| Coordination of Services | 8 | 20 |
| Telemedicine Capabilities | 7 | 18 |
| Educational Materials | 14 | 35 |
| Public Discussion | 14 | 35 |
| Genetic Research | 9 | 23 |
| More Genetic Providers | 6 | 15 |
| Services Closer to Families | 3 | 8 |
| More Family Involvement | 0 | 0 |
| More Newborn Screens | 6 | 15 |
| Other | 3 | 8 |

* Percentages may not equal 100% due to rounding and incomplete responses on some surveys.

## Additional Issues to Consider Regarding Genetic Services in Virginia

An open-ended question was asked of those surveyed requesting that they list any other issues regarding genetic services in Virginia that would be important to consider. Responses included an "increased need for education, research, and access for Virginians," a need for "training for doctors in communicating genetic issues to their patients," a need to increase understanding of "cardiac genetics," and efforts to close "the gap between detection and treatment."

Additional stakeholders wished the researchers to know that "these questions are more important now than five years ago and will be much more important in 5-7 years," and wished to praise the "great follow-up" patients currently receive.

### Impact of These Findings

Those included in this survey are individuals whom were identified as having an important stake in the future of genetic services in the Commonwealth of Virginia. They were selected because they were individuals whom have say in the development of policy and regulations regarding genetics, in the determination of coverage for services, and the potential treatment of patients, among others. It is important for these individuals to possess a strong understanding of genetics and the issues surrounding genetic services. Yet, among this group of key stakeholders more than half rated their own knowledge of genetics as below the level of "good." This is similar to the level of genetic literacy seen in studies of similar subgroups of these stakeholders (1-4). A simple question was included on the survey regarding the importance of folic acid in the prevention of birth defects. This is a health issue that has been the focus of many educational programs, but 44% of those who rated their knowledge level as "good" or "excellent" answered the question incorrectly. Therefore, this self-reported knowledge level may or may not reflect their actual understanding of the topic.

Though 86% of respondents indicated that providing genetic services to Virginian citizens is important, more than 50% indicated a lack of understanding of the current system of genetic services and were uninformed about current accessibility issues, geographic distribution and number of providers, and coverage and protection for genetic services. This is again similar to the level of awareness reported in prior studies (1, 5, and 6). If these stakeholders are individuals who are in a position to play a key role in furthering the development of genetic services, education about the field of genetics and the realities of genetic services will be crucial. This is especially true given that the public often turns to members of these groups for information, the media being a prime source for many. Yet it has been shown that without proper background, media writers often present views that may exaggerate the benefits of certain discoveries while ignoring a complete description of the risks (7-9).

This need for genetic education was the most frequently listed future need for Virginia, closely followed by a need for additional educational materials for families, and public discussion or education about the ethical, social, and legal issues surrounding genetics. Having these three needs so highly ranked is not surprising, seeing as 78% of those asked felt that the average Virginian had a "poor" or "very poor" knowledge of genetics. This continuing theme of a need for education mirrors the findings consistently reported in the other sections of this needs assessment.

# 15. TOWARD DEVELOPING A STRATEGIC STATE GENETIC PLAN: CONCLUSIONS AND RECOMMENDATIONS

This comprehensive needs assessment was initiated to assure that a high quality system of care and interventions are available so that all Virginians can access appropriate services for genetic health concerns. Having a thorough understanding of the current system and where gaps may exist is necessary if Virginia is to develop and sustain the infrastructure required to meet the growing challenges of genetic medicine. This project aimed to describe the current status of genetic services and education in the state and the barriers to high quality care; to assess the current and emerging practices and gaps in services; and to determine ways to address the existing system and the growing disparities in access to genetic services. With the input of the many groups described within this report, four main areas of focus emerged.

## Education

Advances in genetics have opened the door to treatment and the health management of many individuals with identified genetic conditions. Taking full advantage of these advances requires an awareness of the resources, a thorough understanding of what genetics can and can not do, as well as an understanding of the risks, benefits, and limitations of genetic diagnosis and testing. Yet, each group queried during this assessment identified a lack of understanding or knowledge of genetics as one of the biggest challenges facing the state. A need to develop an education program for the general public, stakeholders, as well as healthcare providers was listed as a top priority.

Recommendations to consider in the development of an education plan include:

- Involve the community in planning to determine what information is needed and wanted. Planning must take into account the public's perceptions of genetics and genetic services.
- Collaborate with the March of Dimes and other non-profit organizations to help promote genetic education.
- Develop partnerships with the media to assure accurate reporting on genetic topics.
- Focus on increasing the awareness of ethical, legal, and social issues surrounding genetics.
- Promote efforts to incorporate formal genetic education into all phases of education including K-12.
- Work to dispel myths and misconceptions.
- Explore cost effective methods of education for healthcare providers and students in training.
- Provide instruction to primary care providers on how to address genetic questions, as they are often where the public turns for answers.
- Demonstrate to primary care providers the relevance of genetic issues to their current practice.
- Develop additional education materials for families and providers.
- Collaborate with other states that have or are developing an education plan.

A formal education program for the state should lead to more informed citizens and providers. This increase in genetic literacy is necessary to maximize the value of genetic services and should lead to an improvement in health and quality of life.

## Provision of Genetic Services

Though the quality of the current genetic services offered in Virginia was consistently rated highly by participants, a number of barriers to the accessibility of these services were identified. These included geographic barriers, with limited services available in some portions of the state, particularly rural areas; an insufficient number of genetic providers; and cultural and linguistic challenges that have yet to be addressed.

Recommendations to address these barriers and improve genetic services include:

- Promote and enhance the visibility of the currently available genetic services in Virginia.
- Work to increase the availability of cancer genetic services and adult genetic services.
- Collaborate with health care providers and local and state programs to improve awareness of genetic services and identify individuals in need of referral.
- Coordinate healthcare systems and collaborate with other providers to ensure continuity of care for individuals with genetic conditions. Consider multidisciplinary meetings and providing patients with specialist referrals.
- Consider use of non-physician providers in underserved areas of the state.
- Recruit additional genetic providers.
- Establish additional regional satellite clinics.
- Promote early detection to allow benefit from early treatment or prevention options.
- Continue to assess methods of utilizing current databases to identify individuals and families who may benefit from genetic services.
- Investigate the increased use of telemedicine to serve rural areas.
- Establish quality assurance guidelines for genetic providers, including standard of care guidelines.
- Establish referral guidelines for non-genetic healthcare providers.
- Increase the use of accurate family histories by healthcare providers to identify individuals in need of genetic services.
- Work to improve the risk assessment skills of healthcare providers to assure appropriate referrals.
- Work to assure that healthcare providers and the Virginia public health system meet the genetic core competencies outlined by NCHPEG and the CDC to assure quality care.
- Translate educational materials to benefit the culturally diverse population served in Virginia.

Addressing the barriers noted by the participants and making improvements to our current system of genetic services will assure that those in need are identified and have access to care.

### Funding Issues

Genetic services have been shown to have an impact on the health and quality of life of individuals and their families. Genetic services can lead to prevention, early detection, and early treatment. The participants in this needs assessment overwhelmingly expressed that the availability of these services to the citizens of Virginia is vital and important. However, funding issues have been identified as a major impediment to the availability of these genetic services. Clinical genetic care is very time-consuming, cognitively labor-intensive, and emphasizes preventative healthcare. It has historically not been well reimbursed by insurers. Though many insurance plans may now cover a genetic consultation, they often have strict limitations on coverage, if any, for genetic testing. This limited insurance coverage and resulting high cost of genetic services prevents a significant portion of patients from receiving valuable care. Improving the funding of these genetic services needs to be a major focus for the future.

Recommendations for improving the funding of genetic services include:

- Encourage funding for genetic services through state appropriations and grants.
- Work to increase reimbursement from insurance companies for genetic services and testing.
- Expand state-supported genetic services.
- Educate insurers and third-party payers about genetic issues.
- Increase communication and partnerships with insurers and third-party payers.
- Assist and participate in policy development, including expanded benefit coverage.
- Determine methods of demonstrating the cost-effectiveness of genetic services.

- Collaborate with national organizations addressing funding issues.
- Promote relevant insurance legislation.
- Increase the role VDH plays in reimbursement issues.

Addressing these funding issues will help to improve the accessibility of genetic services to the citizens. It will help to assure that those with genetic conditions receive the care they need and deserve without financial concern.

### Discrimination Issues

The possibility that an individual's genetic information may be used against them in decisions regarding their employability and insurability was an issue of concern raised by a variety of groups during this needs assessment. Virginia does have state laws that place some protections against discrimination in employment and health insurance, yet there is still little protection against discrimination in life insurance, disability, and long-term care insurance. Many participants felt that the current protection was insufficient or they lacked an understanding of the laws currently in place. A fear of discrimination may prevent individuals from pursuing needed services.

Recommendations for addressing discrimination issues include:
- Educate the public and healthcare providers about the current Virginia laws and their protections.
- Monitor and provide comment to state and federal authorities regarding discrimination laws.
- Advocate for improvements to state laws that protect against genetic discrimination.
- Advocate for improvements to state laws that protect and regulate the sharing of genetic information.
- Educate non-genetic providers about practice patterns that will protect genetic privacy and informed consent.

Though the consumers in this needs assessment reported that they had not personally experienced any discrimination as a result of pursuing genetic services, and the genetic providers report they are unaware of any cases of discrimination among patients in their practice, a fear does still remain. This fear, whether real or perceived, may prevent individuals from getting valuable care. As a result, it needs to be addressed.

### State Genetic Plan

With the insights obtained from this needs assessment process, the next step should include the development of a comprehensive state genetic plan that will meet the needs of Virginia. Formalized state plans have been established in 19 or more states and have provided a detailed program to improve genetic healthcare within these states. Virginia's state plan which would encompass the opinions of healthcare providers, policymakers, consumers, key stakeholders, and the general public will allow health promotion efforts to be more effective, will help more people gain access to quality genetic services, and ensure that personal genetic information is protected. The establishment of a state plan will allow for a true integration of genetics into healthcare.

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# APPENDICES

## Appendix A. Genetic Services Assessment

### *RESEARCH SUBJECT INFORMATION AND CONSENT FORM*

**TITLE:** 2003 Virginia GeneSEAN: Genetic Services and Education – Assessment of Needs
(Genetic Provider Survey)

**VCU IRB NO.:** 3291

**SPONSOR:** March of Dimes, Virginia Chapter

**Purpose of the Study:**
The purpose of this research study is to determine the current status of medical genetic services and education in Virginia with regard to existing systems of care, prevention services, educational activities, insurance issues, data collection, quality assurance and ongoing evaluation, as well as gaps in these systems. You are being asked to participate in this study because you have been identified as a clinical genetics professional/provider practicing in the Commonwealth of Virginia.

**Description of the Study and Your Involvement:**
If you decide to be in this research study, complete the enclosed survey only after you understand what is involved and have had all of your questions answered. In this study you will be asked to complete a survey detailing your activities as a clinical genetics provider and your views on critical issues affecting the profession.

**Risks and Discomforts:**
There are no known risks or discomforts involved with participation in this study.

**Benefits:**
There may be no direct benefits to you resulting from participation in this study, however, the resulting data may help improve genetic services and education in Virginia in the future.

**Costs:**
It is anticipated that completion of the survey will take approximately 30-45 minutes of your time.

**Alternatives:**
You have the option not to participate in this study.

**Confidentiality:**
Your responses will be confidential and the documents and publications developed from the survey data will not identify individual respondents. All findings will be presented in aggregate form.

**Voluntary Participation and Withdrawal:**
Participation in this study is voluntary. You may also choose not to answer particular questions that are asked in the survey.

**Questions:**
If you have any questions about the study or your participation, contact Dr. Joann Bodurtha at 804-828-9632 or bodurtha@hsc.vcu.edu. If you have any questions about your rights as a participant in this study, you may contact:
Office for Research Subjects Protection
Virginia Commonwealth University
1101 E. Marshall St., Room 1-023
P.O. Box 980568
Richmond, VA 23298
Telephone: 804-828-0868

**Consent:**
If you understand this consent, have no questions about the study and agree to participate, no signature is required. Your completion and submission of the survey will serve as your consent. If you agree to participate, please complete and return the survey.

# *GENETIC PROVIDER SURVEY*

*Thank you for participating in our survey. Your responses are valued and important. The survey should take 30-45 minutes to complete. Please indicate which is the most appropriate answer for each question. If a question is not applicable to you, indicate NA. If you don't know a particular answer, indicate DK. We would also appreciate your input in the form of patient examples or "stories," so if you know of a compelling story that illustrates an issue addressed in one of the questions, please tell us about it in the "Comments" section at the end. Please do not use real names or any other details that could be linked to a particular patient in any of the examples or stories you share with us.*

1. Gender:
a) male b) female

2. Race/ethnicity:
a) White b) Black/African Am., or Negro
c) American Indian/Alaska Native d) Asian
e) Native Hawaiian/other Pacific Islander f) Spanish/Hispanic/Latino
g) other, specify: _____

3. Professional degree(s):
a) M.S./M.A. b) M.P.H. c) D.O. d) M.D. e) Ph.D. f) other: _____

4. Professional certification(s); please indicate whether board certified (BC) or eligible (BE):
____a) Genetic Counseling ____b) Clinical Molecular Genetics
____c) Medical Genetics ____d) Clinical Cytogenetics
____e) Ph.D. Medical Genetics ____f) Molecular Genetic Pathology
____g) Clinical Biochemical Genetics ____h) Other (specify): _____

5. How many years have you worked in the genetics profession?
a) <5 b) 5-10 c) 11-15 d) 16-20 e) >20

6. Do you currently work full-time in the genetics profession? If not, specify hours per week.
a) Yes b) No: ____ hours per week

7. Please indicate which best describes your primary workplace setting:
a) Academic medical center/university hospital b) Commercial laboratory
c) HMO/managed care organization d) Pharmaceutical/biotech company
e) Hospital f) Government agency (non-military)
g) Medical practice – single specialty h) U.S. military
i) Medical practice – multiple specialty j) Other (specify): _____

8. In which city or county is your primary workplace located? _____

9. Please indicate the type(s) of clinic(s) in which you see genetic patients:
a) prenatal b) pediatric c) general d) cancer e) multidisciplinary f) other: _____

10. Please indicate the type(s) of services your group provides:
a) specimen collection for outside or in-house lab testing b) support services

c) individual and family genetic counseling
d) inpatient consultation
e) medical management of genetic conditions
f) genetic autopsy
g) nutritional metabolic management
h) care coordination
i) other (specify): _____

11. If you checked "specimen collection for outside or in-house lab testing" above, please indicate the categories of testing/screening for which your group provides services:

a) preconception carrier screening
b) adult-onset disease testing
c) prenatal screening/diagnosis
d) cancer genetic testing
e) newborn/child diagnostic testing
f) other (specify): _____

12. How many hours/week do you typically spend in each of the following genetic activities?

_____direct patient care (not including patient-related administration)
_____patient-related administration
_____non-patient-related administration
_____teaching
_____study
_____research
_____other (specify) _____

13. What data does your program collect about the patients served and services provided, including, but not limited to, number of patients seen, diagnosis/indication, place of residence, genetic testing performed, referral origin, type of insurance, etc.?

14. How many of each patient type did you personally see in each of the last five years? (Please fill out as much as possible).

| | Prenatal genetics | General genetics | Metabolic genetics | Cancer genetics | Other (specify) _____ |
|---|---|---|---|---|---|
| 1998 new | _____ | _____ | _____ | _____ | _____ |
| 1998 follow-up | _____ | _____ | _____ | _____ | _____ |
| 1999 new | _____ | _____ | _____ | _____ | _____ |
| 1999 follow-up | _____ | _____ | _____ | _____ | _____ |
| 2000 new | _____ | _____ | _____ | _____ | _____ |
| 2000 follow-up | _____ | _____ | _____ | _____ | _____ |
| 2001 new | _____ | _____ | _____ | _____ | _____ |
| 2001 follow-up | _____ | _____ | _____ | _____ | _____ |
| 2002 new | _____ | _____ | _____ | _____ | _____ |
| 2002 follow-up | _____ | _____ | _____ | _____ | _____ |
| 01-06/2003 new | _____ | _____ | _____ | _____ | _____ |
| 01-06/2003 follow-up | _____ | _____ | _____ | _____ | _____ |

15. If you are the director of clinical genetic services at your center, please indicate how many of each patient type were seen at your center in each of the last five years.

| | Prenatal genetics | General genetics | Metabolic genetics | Cancer genetics | Other (specify) _____ |
|---|---|---|---|---|---|
| 1998 new | _____ | _____ | _____ | _____ | _____ |
| 1998 follow-up | _____ | _____ | _____ | _____ | _____ |
| 1999 new | _____ | _____ | _____ | _____ | _____ |
| 1999 follow-up | _____ | _____ | _____ | _____ | _____ |

| | | | | | |
|---|---|---|---|---|---|
| 2000 new | _____ | _____ | _____ | _____ | _____ |
| 2000 follow-up | _____ | _____ | _____ | _____ | _____ |
| 2001 new | _____ | _____ | _____ | _____ | _____ |
| 2001 follow-up | _____ | _____ | _____ | _____ | _____ |
| 2002 new | _____ | _____ | _____ | _____ | _____ |
| 2002 follow-up | _____ | _____ | _____ | _____ | _____ |
| 01-06/2003 new | _____ | _____ | _____ | _____ | _____ |
| 01-06/2003 follow-up | _____ | _____ | _____ | _____ | _____ |

16. Please indicate who, if anyone, supervises your activities as a clinical genetics provider:

a) Medical geneticist b) Ob-gyn physician
c) MFM physician d) Genetic counselor
e) None f) Other (specify): _____

17. In what cities/counties do you participate in outreach genetics clinics (i.e. not including clinics held at your main location)?

18. Please indicate how frequently each of the outreach genetics clinics listed above are held and what patient types are seen at each.

19. How often do you use teratogen information resources in your practice?

a) never b) 1/month c) 1/week d) 1/day e) >1/day

20. What teratogen information resources do you use?

21. Please indicate your level of agreement: A teratogen information hotline in Virginia would be a good use of resources.

a) strongly disagree b) disagree c) neutral d) agree e) strongly agree

22. How often do you use educational materials with your patients?

a) rarely b) sometimes c) often d) usually e) always

23. What educational materials do you generally use?

24. Do you think you have sufficient educational materials for your patients? If not, why?

a) yes b) no c) not sure

25. Do you think you have sufficient resources within your program/institution to provide culturally and linguistically appropriate patient care?

a) yes b) no c) not sure

26. What deficiencies or barriers within your program, if any, hinder the provision of culturally and linguistically appropriate care?

27. How many of your patients have a first language other than English?
a) < 1% b) 1-4% c) 5-9% d) 10-14% e) ≥15%

28. Please list the first languages, other than English, of your patients.

29. Resources regarding which culture(s) and language(s) are needed and lacking within your program?

30. What is your perception of the overall quality of clinical genetic services in Virginia?
a) very poor b) poor c) neutral d) good e) excellent

31. Please indicate your level of agreement: There are problems with the accessibility or availability of clinical genetic services in Virginia. (Please describe these problems, if any, below.)
a) strongly disagree b) disagree c) neutral d) agree e) strongly agree

32. Please indicate your level of agreement: The number of genetic providers (clinical geneticists, genetic counselors, genetic nurses, metabolic nutritionists, etc.) practicing in Virginia is sufficient to meet current needs.
a) strongly disagree b) disagree c) neutral d) agree e) strongly agree

33. Please indicate your level of agreement: The geographic distribution of genetic providers in Virginia is sufficient to meet current needs.
a) strongly disagree b) disagree c) neutral d) agree e) strongly agree

34. At your center, how far ahead are clinics booked on average (how long do patients usually have to wait to be seen)? Please give a separate response for each applicable clinic type.

Prenatal genetics _____ General genetics _____
Metabolic genetics _____ Cancer genetics _____
Other (specify) _____

35. How do you think access to genetic services in Virginia could be improved?

36. What performance measures or quality assessment activities are used within your program?

37. How often are these activities performed?

38. What performance measures or quality assessment activities do you find particularly informative for improving clinical genetic services?

39. Please indicate your level of agreement: Genetic service providers should follow standard-of-care guidelines.
a) strongly disagree b) disagree c) neutral d) agree e) strongly agree

40. Do you feel you are able to stay up-to-date in genetics?
a) yes b) no c) not sure

41. How often do your patients have problems accessing medical specialists? Please indicate why and provide examples if possible.
a) rarely b) sometimes c) often d) usually e) always

42. How often do your patients have problems obtaining medications (including medical food) or equipment for their condition? Please indicate why and provide examples if possible.
a) rarely b) sometimes c) often d) usually e) always

43. How often do your patients have problems obtaining recommended laboratory testing? Please indicate why and provide examples if possible.
a) rarely b) sometimes c) often d) usually e) always

44. How often do you send labwork to laboratories that you believe are ***not*** the best because of insurance requirements?
a) rarely b) sometimes c) often d) usually e) always

45. How often has this adversely affected patient care? Please provide examples if possible.
a) rarely b) sometimes c) often d) usually e) always

46. How adequate is private insurance reimbursement for your clinical genetics service?
a) very inadequate b) somewhat inadequate c) neutral d) somewhat adequate e) very adequate

47. How adequate is Medicaid/Medicaid HMO reimbursement for your genetics service?
a) very inadequate b) somewhat inadequate c) neutral d) somewhat adequate e) very adequate

48. How does this compare to other services at your institution?
a) much worse b) worse c) similar d) better e) much better

49. [For physician providers only] Approximately how many RVU's (relative value units) do you bill annually?

50. What is the average level of reimbursement received from third party payors relative to *charges billed* for clinical genetic services at your institution?
a) 0-20% b) 21-40% c) 41-60% d) 61-80% e) 81-100%

51. What is the average level of reimbursement received from third party payors relative to the *actual cost* of providing clinical genetic services at your institution?
a) 0-20% b) 21-40% c) 41-60% d) 61-80% e) 81-100%
52. What strategies have you tried to address reimbursement problems?

53. Which of these strategies have been most successful?

54. How significant are insurance referral and authorization problems for your genetics program?
a) very insignificant b) somewhat insignificant c) neutral d) somewhat significant e) very significant

55. Which services are generally authorized and which are not?

56. How much time each week do you personally spend trying to obtain coverage/reimbursement for genetics services from third party payors?

57. What funding sources for clinical genetics services are available at your institution, other than insurance reimbursement?

58. How do you think the billing and reimbursement issues facing genetics services in Virginia should be addressed?

59. How often are your patients concerned about genetic discrimination and privacy issues?
a) rarely b) sometimes c) often d) usually e) always

60. As far as you are aware, how many of your patients have experienced genetic discrimination? Please provide examples if possible.
a) none b) <5% c) 5-9% d) 10-20% e) >20%

61. How satisfied are you with the level of protection current genetic discrimination and privacy laws provide for Virginians?
a) very dissatisfied b) dissatisfied c) neutral d) satisfied e) very satisfied

62. What role(s) do you think the Virginia Department of Health and other statewide genetic leadership organizations ***currently*** play with regard to clinical genetic services in Virginia?

63. What role(s) do you think the Virginia Department of Health or other statewide genetic leadership organizations ***should*** play with regard to clinical genetic services in Virginia?

64. How important do you think having a "medical home" (provider who manages the patient's medical care) is for your patients?
a) very unimportant b) somewhat unimportant c) neutral d) somewhat important e) very important

65. How many of your patients have a "medical home"?
a) <10% b) 10-24% c) 25-49% d) 50-75% e) >75%

66. ***To*** which specialties/agencies do you regularly refer patients (or recommend PCP refer to)?

67. ***From*** which specialties/agencies do you regularly receive referrals?

68. How often do you refer patients to or enroll patients in genetic research protocols?

a) rarely b) sometimes c) often d) usually e) always

69. What percentage of referrals for clinical genetic services in Virginia are made appropriately? Please elaborate.
a) <20% b) 20-39% c) 40-59% d) 60-80% e) >80%

70. How do you think referral appropriateness could be improved?

71. What specialties/agencies, if any, do you think particularly need education about appropriate genetics referrals?

72. How satisfied are you with Virginia's non-genetic healthcare providers' level of knowledge and understanding of genetics?
a) very dissatisfied b) dissatisfied c) neutral d) satisfied e) very satisfied

73. How satisfied are you with Virginia's non-genetic healthcare providers' level of knowledge and understanding of clinical genetic services?
a) very dissatisfied b) dissatisfied c) neutral d) satisfied e) very satisfied

74. In your experience, how often is the training of the general medical community in Virginia adequate to provide appropriate care for their patients with birth defects and genetic conditions?
a) rarely b) sometimes c) often d) usually e) always

75. How often are you involved with the genetic education of non-genetic healthcare providers in Virginia?
a) $\leq$ once/year b) 2-5/year c) 6-11/year d) 1-2/month e) $\geq$ 3/month

76. Please list the types of educational genetic activities for providers with which you are involved and the specialties of the providers targeted.

77. How satisfied are you with the level of Virginians' knowledge and understanding of genetics?
a) very dissatisfied b) dissatisfied c) neutral d) satisfied e) very satisfied

78. How satisfied are you with the level of Virginians' knowledge and understanding of clinical genetic services?
a) very dissatisfied b) dissatisfied c) neutral d) satisfied e) very satisfied

79. How often are you involved with the genetic education of consumer groups or the public in Virginia?

a) ≤ once/year b) 2-5/year c) 6-11/year d) 1-2/month e) ≥ 3/month

80. Please list the types of educational genetic activities targeting the public with which you are involved.

81. What do you think are the top three deficiencies or barriers facing clinical genetic services in Virginia?
82. What do you think are the top three successes of clinical genetic services in Virginia?

83. Additional comments:

*Thank you for your input. Your responses will be confidential and information from the survey will not identify individual respondents, however, we would like to contact you if any clarification or elaboration of your responses is needed. If this is acceptable to you, please indicate your contact information below. Once clarifications are obtained, all identifying information will be removed. If you have any questions or concerns, please feel free to contact Joann Bodurtha at 804-828-9632 (phone), 804-828-3760 (fax) or bodurtha@hsc.vcu.edu, or Katherine Teague at 757-318-6660 (phone), 757-318-6395 (fax) or katherineteague@hotmail.com.*

Name: _____
Telephone: _____
Fax: _____
Email: _____

*Please email, fax or mail the completed survey to:*

*Dr. Joann Bodurtha*
Dept. of Human Genetics
*Virginia Commonwealth University*
*P.O. Box 980033*
*Richmond, VA 23298-0033*

***or***

*Dr. Joann Bodurtha*
Dept. of Human Genetics
*(804) 828-3760 (fax)*
*bodurtha@hsc.vcu.edu*

## Appendix B. Physician Survey

### *COVER LETTER*

June 2004

Dear Physician,

Input from the physicians of Virginia is vital to the shape of healthcare in the Commonwealth. We ask for your help in completing the attached survey on the status of genetic services in our state. This survey is the next part of the Virginia GeneSEAN study: Genetic Services and Education, Assessment of Needs. The study, which is supported by a grant from the March of Dimes, will lead to a document detailing the current status of medical genetic services and education throughout Virginia. We hope you will help us understand better what is going well and where there are gaps.

Your response to the enclosed survey is important. This is an opportunity to share your views on critical issues affecting families with genetic conditions and genetic susceptibilities. Your responses will be confidential and documents developed from the survey data will not identify individual respondents. All findings will be presented in aggregate form. The ultimate goal of this project is to provide a document which is useful to health professionals, educators and policymakers engaging in initiatives to improve the quality of genetic health care in Virginia.

We greatly appreciate your willingness to complete this survey. Please fax or mail the completed survey to Elena Zvirbulis. If you have any questions or comments please feel free to contact us. Thank you for your time and interest.

Sincerely,

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## *Physician Survey*

1) How do you identify family history of a genetic condition in your practice? (Check all that apply)
__ patients fill out standard forms at initial visit
__ I ask patients about family history if they express a concern about it
__ I ask patients about family history if it is relevant to their presenting medical condition
__ I ask patients questions about their family history to identify conditions for which they may be at risk
__ I refer family history questions to genetic counselors / geneticists
__ I rarely ask patients about their family history
__ other

2) How do you usually address family / patient questions about genetic issues?
__ I handle most of these questions within our practice
__ I refer most of these patients to genetic counselors / geneticists
__ I refer most of these patients to specialists other than genetic counselors / geneticists
__ other (please specify) _____

3) Which of the following types of patients do you refer to genetic counselors / geneticists? (Check all that apply)
pregnant women with : __ abnormal serum screen, __fetal anomaly found on ultrasound, __ age 35 or over
children with: __ birth defects, __ mental retardation, __identified by newborn screen
patients with: __ metabolic disorders, __ family history of cancer, __ family history of sudden death
other:_____(please specify)

4) With which groups do you discuss folic acid (0.4 mg daily) prevention of neural tube defects? (Check all that apply)
__ women who are pregnant __ teenagers
__ women who are planning a pregnancy __ pamphlets are available
__ women of childbearing age __ do not discuss

5) Approximately how many patients do you see each year? _____

6) Approximately how many patients do you refer to genetic counselors / geneticists each year?
__ none __ 1-20 __ 21-40 __41-60 __ 61-80 __81-100 __more than 100

7) If you have referred patients to genetics, have you been satisfied with genetics services? __ yes __ no

8) Do you think there are sufficient genetics services in Virginia? __ yes __ no

9) Has cost of an appointment prevented patients you refer from receiving genetics services? __ yes __ no

10) Has distance to a genetics clinic prevented patients you refer from receiving genetics services? __ yes __ no

11) What percentage of patients that you refer to genetic counselors / geneticists are seen by these providers?
__100% __90% __80% __70% __60% __50% __40% __30% __20% __10% __0%

12) Please assess current genetic services in Virginia: __excellent __above average __average __below average __poor

13) Please rate your knowledge of genetics from 1 to 10 (1 is lowest, 10 is highest): ____

14) Please rate your need for education in genetics from 1 to 10: ____

15) Please rate physicians in general's need for education in genetics from 1 to 10: ____

16) Please specify for which genetic conditions / issues physicians need additional education:
_____

17) Please rank the 3 best ways physicians' needs for genetics education can be met (1=best, 2= second, 3= third):

__ local education / seminars __ teleconference __ statewide CME conferences
__ journals __ brochures __ newspaper articles
__ websites __ computer CD roms __ videos
__ television commercials __ teach more genetics in medical school

18) Please rate the general population's need for education in genetics from 1 to 10 (1 is lowest, 10 is highest): ____

19) Please specify for which genetic conditions / issues the general public needs additional education:

_____

20) Please rank the 3 best ways the general population's needs for genetics education can be met (1= best, 2= second, 3= third):

__ local education / seminars __ brochures __ newspaper articles
__ websites __ computer CD roms __ videos
__ television commercials __ teach more genetics in high school

21) Please rank the top 3 needs in genetics for Virginians (1= top, 2= second, 3= third):

__ more genetic education __ more genetic research
__ more application of genetic research to clinical problems __ more genetic providers
__ improve coordination of services __ services closer to families, esp. rural
__ telemedicine capabilities for genetic consultation __ more family involvement
__ more educational materials for families __ more newborn screening tests
__ more public discussion of ethical / legal / social issues __ other (please specify)_____

22) Please rank the following roles of the Virginia Department of Health with regard to genetic services in order of importance (1=most important, 8= least important):

__ newborn screening follow-up __ birth defects registry
__ ensuring availability of genetic services __ ensuring access to lab testing
__ planning for the future in state genetics services __ genetics education
__ ensuring that population screening is appropriately introduced __ quality assurance
__ other _____(please specify)

23) What is your gender? __ male __ female

24) What is your race / ethnicity?

__ Caucasian __African American __ Hispanic
__ Asian __ Native American __ Other (please specify) _____

25) What is your medical specialty? (Check all that apply)

__ Family Practitioner __ Internist __ Ob/Gyn __ Pediatrician
__ VDH director __ Other (please specify) _____

26) In what setting do you most frequently practice medicine? __ urban __ suburban __ rural

27) How many years have you worked as a physician?

__ less than 5 __5-10 __11-15 __16-20 __21-25 __26-30 __ 30+

28) Suggestions for improving genetics services: _____

29) Additional comments: _____

Thank you. Please fax to Elena Zvirbulis (804) 828-7094.

March of Dimes
Saving babies, together

## *Core Competencies in Genetics Essential for All Health-Care Professionals*

The National Coalition for Health Professional Education in Genetics (NCHPEG) endorsed these core competencies on 14 February 2000. NCHPEG is an interdisciplinary group comprising leaders from approximately 120 diverse health professional organizations, consumer and voluntary groups, government agencies, private industries, managed-care organizations, and genetics professional societies. NCHPEG is a national effort to promote health-professional education and access to information about advances in human genetics, to improve the nation's health.

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**PURPOSE**
The impetus for developing the ideal competencies related to genetics was to encourage health-care providers to integrate genetics knowledge, skills, and attitudes into routine health care to provide effective care to individuals and families.

The Core Competency and Curriculum Working Group of NCHPEG recommends that all health professionals possess the core competencies in genetics, as identified in this report, to enable them to integrate genetics effectively and responsibly into their current practice.
Competency in these areas represents the minimum knowledge, skills, and attitudes necessary for health professionals from all disciplines (medicine, nursing, allied health, public health, dentistry, psychology, social work, etc.) to provide patient care that involves awareness of genetic issues and concerns.

***Each health care professional should at a minimum be able to:***
Appreciate limitations of his or her genetic expertise.

- Understand the social and psychological implications of genetic services
- Know how and when to make a referral to a genetics professional.

**BACKGROUND**
During the last decade, the evolution of scientific discoveries from the study of genetics has provided information with potential for tremendous influence on health care. Understanding the role genetics plays in health and disease provides the means to integrate such information into diagnosis, prevention, and treatment of many common diseases and to improve the health of society. Genetic discoveries are already making their way into mainstream healthcare. Patients are beginning to ask providers about genetic services. Primary-care professionals face economic, institutional and professional opportunities and challenges in managing persons at risk for inherited conditions. As outlined by the Institute of Medicine Report on the Future of Public Health (IOM, 1988), public health agencies will have an increasing role in assessing the health needs of populations, working with the private sector in ensuring the quality of genetic tests and services, and evaluating the impact of interventions on medical, behavioral, and psychosocial outcomes. Ultimately, health- care providers, regardless of specialty area, role, or practice setting, will face questions about implications of genetics for their patients. The fast pace of genetic advances and the paucity of professional training in genetics leave many providers without up-to-date answers for their patients.

**IMPLEMENTATION**
It is essential that persons and groups responsible for continuing education, curriculum development, licensing, certification, and accreditation bodies for all health-care disciplines adopt these recommendations and integrate

genetics content into ongoing education. The competencies provide direction for curriculum content that can be used in the design of seminars, workshops, and academic preparation. There is a need for commitment on the part of all educators to incorporate genetic information into all levels of professional education. Enhanced genetics competency will help us to meet the changing demands of the health-care system and promote human benefit as a result of discoveries in genetics and genetic medicine. Although this list may appear challenging, it is important to prepare for the reality of tomorrow and not only for the needs of today.

This document is a work in progress, because it is likely that the knowledge produced by the Human Genome Project and related activities will create an ongoing need to assess and revise expectations. Although the list is extensive, NCHPEG believes that the recommendations provide a useful tool for organizing the teaching of basic genetics in many educational settings and can be modified for a particular discipline.

Those health professionals involved in the direct provision of genetics services may require additional training to achieve an appropriately higher level of competence. Indeed, there are a number of examples of specific recommendations for training of professionals who require specialized knowledge of genetics (AAFP, 1999; ASCO, 1997; ASHG, 1995; APHMG, 1998; Fine et al, 1996; Hayflick & Eiff, 1998; Jenkins, 2000; Stephenson, 1998; & Taylor-Brown & Johnson, 1998).

**RECOMMENDATIONS**

Note: Throughout this document, the term "clients" includes individuals and their sociological and biological families.

**KNOWLEDGE**

*All health professionals should understand:*

1.1 basic human genetics terminology
1.2 the basic patterns of biological inheritance and variation, both within families and within populations
1.3 how identification of disease-associated genetic variations facilitates development of prevention, diagnosis, and treatment options
1.4 the importance of family history (minimum three generations) in assessing predisposition to disease
1.5 the role of genetic factors in maintaining health and preventing disease
1.6 the difference between clinical diagnosis of disease and identification of genetic predisposition to disease (genetic variation is not strictly correlated with disease manifestation)
1.7 the role of behavioral, social, and environmental factors (lifestyle, socioeconomic factors, pollutants, etc.) to modify or influence genetics in the manifestation of disease
1.8 the influence of ethnoculture and economics in the prevalence and diagnosis of genetic disease
1.9 the influence of ethnicity, culture, related health beliefs, and economics in the clients' ability to use genetic information and services
1.10 the potential physical and/or psychosocial benefits, limitations, and risks of genetic information for individuals, family members, and communities
1.11 the range of genetic approaches to treatment of disease (prevention, pharmacogenomics/prescription of drugs to match individual genetic profiles, gene-based drugs, gene therapy)
1.12 the resources available to assist clients seeking genetic information or services, including the types of genetics professionals available and their diverse responsibilities
1.13 the components of the genetic-counseling process and the indications for referral to genetic specialists
1.14 the indications for genetic testing and/or gene-based interventions
1.15 the ethical, legal and social issues related to genetic testing and recording of

genetic information (e.g., privacy, the potential for genetic discrimination in health insurance and employment)

1.16 the history of misuse of human genetic information (eugenics)

1.17 one's own professional role in the referral to genetics services, or provision, follow-up, and quality review of genetic services

**SKILLS**

*All health professionals should be able to:*

2.1 gather genetic family-history information, including an appropriate multi-generational family history

2.2 identify clients who would benefit from genetic services

2.3 explain basic concepts of probability and disease susceptibility, and the influence of genetic factors in maintenance of health and development of disease

2.4 seek assistance from and refer to appropriate genetics experts and peer support resources

2.5 obtain credible, current information about genetics, for self, clients, and colleagues

2.6 use effectively new information technologies to obtain current information about genetics

2.7 educate others about client-focused policy issues

2.8 participate in professional and public education about genetics

*Skills 2.9-2.17 delineate the components of the genetic-counseling process and are not expected of all health-care professionals. However, health professionals should be able to facilitate the genetic-counseling process and prepare clients and families for what to expect, communicate relevant information to the genetics team, and follow up with the client after genetics services have been provided. For those health professionals who choose to provide genetic-counseling services to their clients, all components of the process, as delineated in 2.9-2.17 should be performed.*

2.9 educate clients about availability of genetic testing and/or treatment for conditions seen frequently in practice

2.10 provide appropriate information about the potential risks, benefits, and limitations of genetic testing

2.11 provide clients with an appropriate informed consent process to facilitate decision making related to genetic testing

2.12 provide, and encourage use of, culturally appropriate, user-friendly materials/media to convey information about genetic concepts

2.13 educate clients about the range of emotional effects they and/or family members may experience as a result of receiving genetic information

2.14 explain potential physical and psychosocial benefits and limitations of gene-based therapeutics for clients

2.15 discuss costs of genetic services, benefits and potential risks of using health insurance for payment of genetic services, potential risks of discrimination

2.16 safeguard privacy and confidentiality of genetic information of clients to the extent possible

2.17 inform clients of potential limitations to maintaining privacy and confidentiality of genetic information

**ATTITUDES**

*All health professionals should:*

3.1 recognize philosophical, theological, cultural, and ethical perspectives influencing use of genetic information and services

3.2 appreciate the sensitivity of genetic information and the need for privacy and confidentiality

3.3 recognize the importance of delivering genetic education and counseling fairly, accurately, and without coercion or personal bias

3.4 appreciate the importance of sensitivity in tailoring information and services to clients' culture, knowledge and language level

3.5 seek coordination and collaboration with interdisciplinary team of health professionals
3.6 speak out on issues that undermine clients' rights to informed decision making and voluntary action
3.7 recognize the limitations of their own genetics expertise
3.8 demonstrate willingness to update genetics knowledge at frequent intervals
3.9 recognize when personal values and biases with regard to ethical, social, cultural, religious, and ethnic issues may affect or interfere with care provided to clients
3.10 support client-focused policies

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- Agency for Healthcare Research and Quality
- American Academy of Family Physicians
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- American Academy of Oral Medicine
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- American Association for Klinefelter Syndrome Information and Support
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- Association of Family Practice Residency Directors
- Association of Genetic Counseling Program Directors
- Association of Genetic Technologists
- Association of Professional Chaplains
- Association of Professors of Human or Medical Genetics
- Association of Professors of Medicine
- Association of Schools of Allied Health Professions
- Association of Schools of Public Health
- Association of State and Territorial Health Officials
- Association of Teachers of Preventive Medicine
- Association of Womens Health, Obstetric & Neonatal Nurses
- Biotechnology Industry Organization
- Centers for Disease Control and Prevention
- Chicago Center for Jewish Genetic Disorders
- Coalition of State Genetics Coordinators
- College of American Pathologists
- Council of Medical Specialty Societies
- Council on Social Work Education
- Dana Alliance for Brain Initiatives
- Department of Veterans Affairs
- Federation of Special Care Dentistry
- Foundation for Blood Research
- Genetic Alliance
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- Health Resources and Services Administration
- Hoffmann-La Roche
- Howard Hughes Medical Institute
- Human Genome Education Model Project
- International Patient Advocacy Association
- International Society of Nurses in Genetics
- Joint Commission on Accrediation of Health Care Organizations
- March of Dimes
- National Association of Catholic Chaplains
- National Association of Neonatal Nurses
- National Association of Pediatric Nurse Practitioners
- National Association of School Nurses
- National Association of Social Workers
- National Board of Medical Examiners
- National Cancer Institute
- National Center for Genome Resources
- National Heart, Lung, and Blood Institute
- National Human Genome Research Institute
- National Institute of Child Health & Human Development
- National Institute of Dental and Craniofacial Research
- National Institute of Diabetes and Digestive and Kidney Diseases
- National Institute of Environmental Health Sciences
- National Institute of General Medical Science
- National Institute of Mental Health
- National Institute of Neurological Disorders and Stroke
- National Institute of Nursing Research
- National Institute on Aging
- National Institute on Alcohol Abuse & Alcoholism
- National Institute on Deafness and Other Communication Disorders
- National Institute on Drug Abuse
- National Marfan Foundation
- National Medical Association
- National Organization for Rare Disorders
- National Organization of Nurse Practitioner Faculties
- National Society of Genetic Counselors
- Nursing Organization Liaison Forum
- Office of Rare Diseases
- Office on Womens Health
- Oncology Nursing Society
- Pharmaceutical Research and Manufacturers of America
- RSi Communications Group
- Scoliosis Association, Inc.
- Sigma Theta Tau International
- Society for Academic Continuing Medical Education
- Society for Inherited Metabolic Disorders
- Society of General Internal Medicine
- Society of Gynecologic Oncologists
- Society of Pediatric Nurses
- Society of Teachers of Family Medicine
- SREB Council on Collegiate education for Nursing

- Tourette Syndrome Association, Inc.
- Uniformed Services Academy of Family Physicians (18)

*December 27, 2001*

# Association of Professors of Human and Medical Genetics/ American Society of Human Genetics MEDICAL SCHOOL CORE CURRICULUM IN GENETICS

***PREAMBLE***

- Medical genetics is one of the most rapidly advancing fields of medicine, and molecular genetics is now integral to all aspects of biomedical science. Every physician who practices in the 21st century must have an in-depth knowledge of the principles of human genetics and their application to a wide variety of clinical problems. The American Society of Human Genetics and the Association of Professors of Human and Medical Genetics have developed this Medical School Core Curriculum to provide guidance to deans and curriculum committees regarding medical genetics knowledge, skills, and behaviors that all current medical students will need during their careers as physicians. Each medical school must find the best way to incorporate genetics teaching into its own curriculum, but some generalizations are possible:
- Medical genetics provides a unique perspective on function of the human body in health and disease; it is both a clinical specialty and a basic science. Medical genetics teaching must span the entire undergraduate medical school curriculum and continue into the postgraduate years.
- Medical genetics must be explicitly included in the curriculum. Although some aspects of medical genetics overlap with and may be taught by other disciplines, specific learning objectives in medical genetics need to be established.
- A well-qualified medical genetics specialist (or small committee of medical geneticists) should be given the authority and responsibility for implementing the genetics curriculum at each medical school. This responsibility should extend throughout the undergraduate medical curriculum and include involvement in all courses that deal with genetic principles or disorders.
- Medical genetics can be taught effectively by a variety of methods and in various formats. Problem-based learning is particularly well-suited to medical genetics because it involves integration of skills and knowledge from many fields. Genetics can also be taught in various clinical contexts and at different points in clinical training, depending on the particular circumstances at each school. Specific clinical examples are important, but the focus of the curriculum must be on medical genetic principles illustrated by the examples.

Given the rapid advance of medical genetics, this Core Curriculum is a work in progress. (The previous version was published in the American Journal of Human Genetics (1995) 56:535-537.) The American Society of Human Genetics and the Association of Professors of Human and Medical Genetics welcome all comments on these objectives, which will be revised as necessary to reflect changes that occur in our understanding of genetics and its application to medicine.

## *GENERAL MEDICAL COMPETENCIES ESSENTIAL TO MEDICAL GENETICS*

During their training, medical students must acquire many general skills and behaviors that are important in all aspects of clinical practice, including medical genetics. These general competencies include the ability of students to:

1.1 explain the importance of disease prediction and prevention;

1.2 understand the developmental stages of human behavior, maturation, and intelligence;

1.3 apply appropriate techniques for conveying difficult medical information;

1.4 understand how to respond appropriately to patients' defense mechanisms;

1.5 recognize the importance of reiterating information to patients who are anxious or unfamiliar with the concepts being presented;

1.6 recognize the importance of patient confidentiality;

1.7 make appropriate referrals to genetics support groups, community groups, or other resources that can benefit the patient and family;

1.8 respect the autonomy of all patients, but also provide guidance with decision-making when requested;

1.9 respect patients' religious, cultural, social, and ethical beliefs, even if they differ from their own beliefs;

1.10 interpret their own attitudes toward ethical, social, cultural, religious and ethnic issues and develop an ability to individualize each patient or family member;

1.11 cope emotionally with patient responses;

1.12 recognize the limitations of their own skills and seek consultation when necessary;

1.13 effectively use resources such as medical textbooks, research articles, and computer-based systems to obtain information necessary for good patient care;

1.14 apply the principles of evidence-based medicine to clinical practice;

1.15 understand how clinical observations can provide insight into human biology and disease pathogenesis and, through research, lead to improvements in health; and

1.16 undertake a program of life-long learning.

***SPECIFIC KNOWLEDGE REQUIREMENTS***

The practice of modern medicine includes recognition of the role of genetic factors in health and disease. Students must know:

**2.1 Structure and Function of Genes and the General Organization of the Human Genome**

2.1.1 what genes are, how they are organized and controlled, what they do, and how they segregate;

2.1.2 how gene expression is affected by differences in coding and non-coding regions, effects of trans-acting factors, and the structure of chromatin;

2.1.3 how protein function is influenced by mRNA and polypeptide processing and interactions;

2.1.4 how gene activity varies during development and in normal and pathological cell function;

2.1.5 what information can and cannot be predicted from the DNA sequence of a gene;

2.1.6 what information can be obtained from measuring RNA or protein levels that cannot be obtained from the DNA sequence alone;

2.1.7how processes such as gene duplication and divergence, exon shuffling, and the activity of transposable elements help to explain genomic variability, redundancy, and plasticity;

**2.2 Genes and Disease**

2.2.1 the patterns of inheritance characteristic of autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive traits;

2.2.2.factors that affect development of the phenotype in single-gene disorders, including modifier genes, and stochastic and pleiotropic effects, which result in variable expressivity and incomplete penetrance;

2.2.3 the clinical manifestations of common mendelian diseases;the basic principles of inborn errors of metabolism and of pharmacogenetic variations and their general clinical manifestations;

2.2.4 the genetic basis of mitochondrial diseases and the expected inheritance patterns for mitochondrial traits;

2.2.5 the nature of mutations and premutations and how they contribute to human variability and disease;

2.2.6 the concepts and clinical importance of genetic imprinting and uniparental disomy;

2.2.7 how polymorphisms, human gene mapping, and gene linkage and association studies are used in medicine;

2.2.8 the multifactorial nature of most human traits, both normal and abnormal, and the principles of multifactorial inheritance;

2.2.9 how genes interact with other genes and with various environmental factors to produce disease, and how amelioration of non-genetic factors can prevent development of disease in a genetically-predisposed

individual;

**2.3 Chromosomes and Chromosomal Abnormalities**

2.3.1 how genes are organized into chromosomes, how chromosomes replicate in mitosis and meiosis, and how they are transmitted from parent to child;

2.3.2 the clinical features of common numerical, structural, and mosaic chromosomal abnormalities;

**2.4 Population Genetics**

2.4.1 how the principles of population genetics account for varying frequencies of particular mutations in populations, the effects of consanguinity, the continuing occurrence of new mutations, and the resistance of gene frequencies to change by medical intervention;

2.4.2 how evolutionary principles can be used to understand human biology and disease;

**2.5 Genetics in Medical Practice**

2.5.1 how knowledge of a patient's genotype can be used to develop a more effective approach to health maintenance, disease prevention, disease diagnosis, and treatment for that particular individual;

2.5.2 common molecular and cytogenetic diagnostic techniques and how they are applied to genetic disorders;

2.5.3 how constitutional and acquired genetic alterations can lead to the development of malignant neoplasms and how identification of these changes can be used in the diagnosis, management and prevention of malignancy;

2.5.4 the potential advantages, limitations, and disadvantages of presymptomatic testing for genetic disease;

2.5.5 the potential advantages, limitations, and disadvantages of predictive testing for genetic disease;

2.5.6 how appropriate applications of genetic medicine can improve public health, and how to determine whether such interventions are warranted in a particular population;

2.5.7 the alternative approaches and goals of screening programs for genetic diseases in newborn infants, pregnant women, and other adults, and the ethical issues involved in justifying each program;

2.5.8 the existence of and justification for screening programs to detect genetic disease, and the difference between screening and more definitive testing;

2.5.9 conventional approaches to treatment of genetic diseases and the general status of gene-based therapies;

2.5.10 what exposures are likely to be teratogenic in humans and how such exposures can be prevented;

2.5.11 how to recognize and classify congenital anomalies and multiple congenital anomaly syndromes;

2.5.12 the purpose of genetic counseling;

2.5.13 when and how to refer individuals with a genetic disease or congenital anomaly to medical genetics specialists, and why referral is beneficial to the patients;

2.5.14 how novel scientific discoveries are evaluated in a clinical context and applied appropriately to the care of patients;

2.5.15 how legal and ethical issues related to genetics affect general medical practice;

2.5.16 how organizational and economic aspects of the health care system affect delivery of clinical genetic services;

2.5.17 what lessons the history of use and misuse of human genetics teach about the proper application of contemporary medical genetic knowledge.

***SPECIFIC SKILLS***

Students must learn to synthesize factual material related to genetic diseases and to use this information to formulate an appropriate plan for diagnostic evaluation and patient management. They need the ability to:

3.1 elicit a comprehensive family medical history, construct an appropriate medical pedigree, and recognize patterns of inheritance and other signs suggestive of genetic disease in the family history;

3.2 recognize features in a patient's medical history, physical examination or laboratory investigations that suggest the presence of genetic disease;

3.3 identify patients with strong inherited predispositions to common diseases and facilitate appropriate assessment of other at-risk family members;

3.4 recognize and classify common congenital anomalies and patterns of anomalies;

3.5 recognize and initiate the evaluation of patients with inborn errors of metabolism;

3.6 interpret the results of common cytogenetic, molecular genetic, and biochemical genetic diagnostic techniques efficiently;

3.7 estimate recurrence risks for mendelian and multifactorial disorders in affected families;

3.8 use the information that a patient has a genetic predisposition for a particular disease to help reduce the risk of developing that disease or deal with it more effectively if it does develop;

3.9 describe appropriate techniques and approaches to providing genetic counseling for commonly-encountered genetic diseases;

3.10 communicate genetic information in a clear and non-directive manner that is suitable for individuals of different educational, socio-economic, ethnic and cultural backgrounds;

3.11 recognize and accept varying cultural, social, and religious attitudes in relation to issues such as contraception, abortion, parenting, and gender roles;

3.12 utilize community support services and agencies, in particular, support groups for genetic diseases, appropriately;

3.13 provide patients with access to diagnostic and predictive tests that are appropriate for the condition in their family and advise patients of the benefits, limitations, and risks of such tests;

3.14 work with a medical genetics specialist to develop a comprehensive plan for the evaluation and management of patients with genetic disease;

3.15 make available to patients with genetic diseases appropriate treatments, including dietary, pharmacological, enzyme-replacement, transplantation, and gene therapies, as well as anticipatory guidance regarding health screening practices specific to the diagnosis;

3.16 appreciate the important role of biomedical research and acquire skills that enable critical analysis of scientific developments.

***SPECIFIC BEHAVIORS***

Students must learn to be sympathetic, nonjudgmental, and non-coercive counselors who recognize their own limitations and seek consultation whenever necessary. Students should:

4.1 present all relevant options fairly, accurately, and non-coercively;

4.2 be aware of the dilemmas posed by confidentiality when relatives are found to be at risk for a serious disease;

4.3 appreciate the implications that information regarding a genetic abnormality can have for a person's self-image, family relationships, and social status and that patients' reactions may differ depending on factors such as gender, age, culture, and education;

4.4 when appropriate, encourage patient participation in medical research provided the patient and/or family is fully informed and understands the risks and benefits of participation in terms of their own disease, treatment, and social context (19).

## ASSOCIATION OF PROFESSORS OF HUMAN OR MEDICAL GENETICS

# Clinical Objectives in Medical Genetics for Undergraduate Medical Students

***June 1998***

## Preamble

The Association of Professors of Human or Medical Genetics developed these objectives to define the knowledge, skills and attitudes in clinical genetics that all medical students should achieve during the clinical phase of their education. These objectives complement those of the ASHG Medical School Core Curriculum in Genetics, which covers both basic science and clinical aspects of medical student education (*Am J Hum Genet 1995;56:535-537*).

We assume that students who are learning clinical medicine already possess an understanding of the basic principles of human genetics. Clinical training provides students with an opportunity to apply their understanding of these principles to the diagnosis, management and prevention of human disease. Learning medical genetics is a process that should continue throughout a physician's career. Medical students should gain cognitive mastery of the objectives listed below during clinical training. Subsequent postgraduate training should provide appropriate experience in the application of these objectives in primary care or a particular medical specialty.

Medical genetics is a recognized clinical specialty as well as an extensive field of knowledge that provides a unique perspective on function of the human body in health and disease. It is, therefore, essential that a well-qualified medical geneticist (or small committee of medical geneticists) be given the authority and responsibility for implementing the clinical genetics curriculum at each medical school. We recognize that curricula, resources and personnel available for teaching medical genetics differ greatly among medical schools. The objectives included here can be taught in various clinical contexts and at different points in clinical training, depending on the particular circumstances at each school.

During their training, medical students must acquire many general skills and attitudes that are important in all aspects of clinical practice, including medical genetics. These general competencies include the ability of students to:

- appreciate the importance of disease prediction and prevention;
- understand the developmental stages of human behavior, maturation, and intelligence;
- recognize the importance of patient confidentiality;
- apply appropriate techniques for conveying difficult medical information;
- recognize and understand how to respond appropriately to patients' defense mechanisms;
- tolerate and encourage reiteration of information because of patient anxiety or unfamiliarity with the concepts being presented;
- respect patients' religious, cultural, social, and ethical beliefs even if they differ from the students' own beliefs;
- respect the autonomy of all patients while giving appropriate consideration to the difficulties certain

handicapping conditions may pose for decision-making;

- make appropriate referrals to community or other resources that can benefit the patient and family;
- interpret their own attitudes toward ethical, social, cultural, religious and ethnic issues and develop an ability to individualize each patient or family member;
- cope emotionally with patient responses;
- recognize the limitations of their own skills and seek consultation when necessary;
- effectively use resources such as medical textbooks, research articles, and computer-based systems to obtain information necessary for good patient care;
- apply the principles of evidence-based medicine to clinical practice;
- understand how clinical observations can provide insight into human biology and disease pathogenesis and, through research, lead to improvements in health; and
- undertake a programme of life-long learning.

Medical genetics is one of the most rapidly advancing areas of medical practice. The knowledge of medical genetics that is necessary for medical practice is likely to be very different in 10 or 20 years than it is now. The Association of Professors welcomes all comments on these objectives, which we shall revise as necessary to reflect changes that occur in our understanding of genetics and its application to clinical medicine.

**Clinical Objectives in Medical Genetics**

**Students should know:**

1.1 how genetic factors predispose to mendelian and multifactorial diseases and the implications of such predispositions for disease diagnosis, treatment and prevention;

1.2 the clinical manifestations of common mendelian diseases;

1.3 the clinical features of common chromosomal aneuploidies and the signs generally associated with other kinds of chromosomal imbalance;

1.4 how constitutional and acquired genetic alterations can lead to the development of malignant neoplasms and how identification of these changes can be used in the diagnosis, management and prevention of malignancy;

1.5 how knowledge of a patient's genotype can be used to develop a more effective approach to health maintenance, disease diagnosis, and treatment for that particular individual;

1.6 the procedures that are generally employed for prenatal genetic diagnosis and the indications for such testing;

1.7 the advantages, limitations, and disadvantages of presymptomatic testing for genetic disease;

1.8 the existence of and justification for screening programs to detect genetic disease, and the difference between screening and more definitive testing;

1.9 the differences in goals and approach among screening programs for genetic diseases in newborn infants, pregnant women, and other adults;

1.10 conventional approaches to treatment of genetic diseases and the general status of gene-based therapies;

1.11 what exposures are likely to be teratogenic in humans and how such exposures can be prevented;

1.12 indications and appropriate methods for referral of individuals with a genetic disease or congenital anomaly to medical genetics specialists;

1.13 how novel scientific discoveries are evaluated in a clinical context and applied appropriately to the care of patients; and

1.14 how legal and ethical issues related to genetics affect general medical practice.

**Students should be able to:**

2.1 elicit a comprehensive family medical history, construct an appropriate medical pedigree, and recognize patterns of inheritance and other signs suggestive of genetic disease in the family history;

2.2 recognize features in a patient's medical history, physical examination or laboratory investigations that suggest the presence of genetic disease;

2.3 identify patients with strong inherited predispositions to common diseases and facilitate appropriate assessment of other at-risk family members;

2.4 recognize and classify common congenital anomalies and patterns of anomalies;

2.5 recognize and initiate the evaluation of patients with inborn errors of metabolism;

2.6 use common cytogenetic, molecular genetic, and biochemical genetic diagnostic techniques efficiently;

2.7 estimate recurrence risks within an affected family for diseases transmitted as uncomplicated mendelian or multifactorial traits;

2.8 use the information that a patient has a genetic predisposition for a particular disease to help him or her reduce the risk of developing that disease or deal with it more effectively if it does develop;

2.9 describe appropriate techniques and approaches to providing genetic counseling for commonly-encountered genetic diseases;

2.10 communicate genetic information in a manner that is suitable for individuals of various different

educational and cultural backgrounds; and

2.11 work with a medical genetics specialist to develop a comprehensive plan for the evaluation and management of patients with genetic disease (20).

# Medical Genetics

*This document has been endorsed by the American Academy of Family Physicians and was developed in cooperation with the American College of Medical Genetics, the Association of Professors of Human and Medical Genetics, the Association of Departments of Family Medicine, the Association of Family Practice Residency Directors and the Society of Teachers of Family Medicine.*

**Attitudes**

The resident should develop attitudes that encompass the following:

A. Recognition of the importance of the family physician, the medical geneticist and the genetics team as collaborators in the evaluation, diagnosis and management of patients referred for genetic consultation.
B. Recognition of the need for sensitivity to the patient's and family's concerns relating to referral for genetic evaluation and diagnosis of a genetic disorder.
C. Recognition of the importance of confidentiality, ethical and legal issues involved in medical genetics.

**Knowledge**

A. Basic principles of human and medical genetics
    1. Genes and chromosomes
    2. Genogram/pedigree
        a. Components
        b. Preparation
        c. Interpretation
    3. Basic Mendelian inheritance patterns (hair/eye color, blood type)
        a. Autosomal dominant
        b. Autosomal recessive
        c. X-linked dominant
        d. X-linked recessive
    4. Non-Mendelian inheritance patterns
        a. Multifactorial
        b. Mitochondrial (MELAS)
        c. Trinucleotide repeats (fragile X syndrome, Huntington's disease)
        d. Imprinting (Prader-Willi syndrome, Angelman's syndrome)
        e. Uniparental disomy (Prader-Willi syndrome, Angelman's syndrome)
B. Ethical and legal considerations/controversies
    1. Screening for genetic abnormalities
    2. Prenatal-preconception testing
    3. Presymptomatic genetic testing (breast cancer genes, Huntington's disease)
    4. Carrier testing for genetic disorders
    5. Confidentiality
    6. Risk assessment
    7. Responsibility to inform
    8. Discrimination issues (insurance coverage, employment)
    9. Informed consent
    10. Paternity determinations
C. Terminology used in medical genetics (mosaicism, incomplete penetrance, variable expressivity, pleomorphic, malformation, deformation, disruption, dysmorphic, minor/major anomaly, homozygote, heterozygote, allele, polymorphism)
D. Laboratory studies and research
    1. Karyotype
    2. Fluorescent in situ hybridization

3. Polymerase chain reaction, sequencing, mutation detection
4. Gene mapping

E. Limitations of genetic testing (polymorphism versus mutation)

F. The genetic implications of common disorders and conditions

1. Chromosomal abnormalities
   a. Trisomy (13, 18, and 21-Down syndrome)
   b. Sex chromosome anomalies (Turner's syndrome, Klinefelter's syndrome)
   c. Translocations, inversions, deletions
   d. Microdeletion syndromes
   e. Cri du chat syndrome
2. Familial variants
   a. Congenital short stature
   b. Delayed-onset puberty
3. Oncology
   a. Colon cancer
   b. Breast cancer
   c. Ovarian cancer
   d. Prostate cancer
   e. Wilms' tumor
   f. Retinoblastoma
   g. Familial ademomatous polyposis
4. Geriatric disorders
   a. Alzheimer's disease
   b. Parkinson's disease
5. Metabolic disorders
   a. Endocrine (diabetes, thyroid)
   b. Amino acids (phenylketonuria, maple syrup urine disease)
   c. Organic acids
   d. Fatty acid oxidation
   e. Energy metabolism
   f. Lysosomal storage (Tay-Sachs disease)
   g. Syndrome "X"
   h. Lipid disorders
   i. Biotinidase deficiency
   j. Growth hormone deficiency
   k. Precocious puberty
   l. $Alpha_1$ -antitrypsin deficiency
6. Skeletal/connective tissue abnormalities
   a. Talipes
   b. Syndactyly
   c. Osteogenesis imperfecta
   d. Achondroplasia
   e. Scoliosis
   f. Ehlers-Danlos syndrome
   g. Marfan syndrome
7. Cardiopulmonary
   a. Congenital heart disease
   b. Cystic fibrosis
   c. Aortic aneurysm
   d. Familial idiopathic cardiomyopathy
   e. Idiopathic hypertrophic septal hypertrophy (subaortic stenosis)

8. Hematologic disorders
    a. Immunoglobulin deficiencies
    b. Hemoglobinopathies
        1. Sickle cell
        2. Thalassemia
    c. Clotting disorders
        1. Hemophilia
        2. von Willebrand's disease
        3. Hypercoagulable disorders
9. Gastrointestinal abnormalities
    a. Esophageal atresia
    b. Pyloric stenosis
    c. Tracheoesophageal
    d. Esophageal "web"
    e. Obesity
10. Neuromuscular disorders
    a. Charcot-Marie-Tooth disease
    b. Myotonic dystrophy
    c. Tourette's syndrome
    d. Benign familial tremor
    e. Muscular dystrophies
11. Neural tube defects
    a. Spina bifida
    b. Syringomyelia
    c. Impact of folate supplementation
12. Craniofacial abnormalities
    a. Cleft lip and palate
    b. Craniosynostosis syndromes
13. Psychiatric disorders
    a. Attention deficit-hyperactivity disorder
    b. Schizophrenia
    c. Addictive personality disorder
    d. Bipolar affective disorder
    e. Associated with genetic disorders
14. Prenatal abnormalities
    a. Prenatal screening
        1. Alpha-fetoprotein
        2. Multiple marker screening
        3. Ultrasound
    b. Carrier screening
        1. Tay-Sachs disease
        2. Canavan's disease
        3. Gaucher's disease
        4. Cystic fibrosis
        5. Hemoglobinopathies
    c. Maternal influence factors
        1. Medications/drugs and chemical exposure
        2. Diabetes
        3. Infections

G. Approach to the dysmorphic child/adult with multiple congenital abnormalities

H. Common questions and misconceptions
    1. Ionizing radiation

2. Magnetic field effects
3. The Human Genome Project
4. Multiple births (twins)
5. Cloning
6. Genetic engineering

**Skills**

A. Preparation of a genogram/pedigree
B. Identification of local community resources for genetic counseling and consultation
C. Identification of pertinent community groups addressing the needs of patients and families with genetically based disorders
D. Basic genetic counseling for family physicians

**Implementation**

The implementation of this curriculum component should take place during the longitudinal learning experiences throughout the 36 months of training. The curricular content should be integrated into the conference schedule and into teaching activities in the family practice center. Relevant resource and patient information materials should be available in the residency library.

**Resources**

National Academy of Sciences. Genetic Screening: Programs, principles, and research. Washington, D.C.: 1975.
Gould RL. Cancer and genetics: answering your patients' questions. Huntington, N.Y.: PRR, 1997.
Doukas DJ. Primary care and the human genome project: Into the breach. Arch Fam Med 1993; 2:1179-83.
Strong C. Tomorrow's prenatal genetic testing. Arch Fam Med 1993; 2: 1187-93.
Rimoin DL, Connor JM, Pyeritz RE. Emery and Rimoin's principles and practice of medical genetics. 3rd ed. New York: Churchill Livingstone, 1997.
Jorde LB, Carey JC, White RL. Medical genetics. St. Louis: Mosby, 1997.
Thompson MW, McInnes RR, Willard HF. Thompson & Thompson Genetics in medicine. 5th ed. Philadelphia; Saunders, 1991.

**Web Sites**

OMIM Home Page. Online Mendelian Inheritance in Man: http://www.ncbi.nlm.nih.gov/Omim

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## Appendix D. Focus Group Project Materials

### *FOCUS GROUP FRAMEWORK*

***Target locations:***

- 3 geographically distinct locations in the state, ideally including one from each category:
    1) urban –Norfolk
    2) suburban –Fredericksburg
    3) rural –UVA outreach clinic (Lynchburg site)

***Target groups:***

- Include patients recruited from prenatal, general/pediatric, and cancer genetic clinics. Exclude patients seen by project staff (i.e. Bodurtha and Zvirbulis).
- Target enrollment of 8-10 people/group. Consider oversampling of 11-12.

***Activity timeline:***

- As soon as IRB approval is received, call chosen clinic sites to request participation, which would involve distributing patient solicitation letters and assistance in locating and reserving an appropriate room/location for the focus group session.
- The flyers instruct interested patients to call Elena (1-800 number) for more information. She provides basic information about the purpose of the sessions (*telephone script*), takes patient contact information, and checks their availability for selected date(s) [*target focus group dates July-August*].
- Elena sends confirmation letters to scheduled participants with details about the location, directions, date and time of their focus group, along with contact information.
- Discontinue patient solicitation letter distribution at a given site when target enrollment is reached.
- Elena calls participants 2-3 days before their session as a reminder.
- Staff team, to include a moderator (Sara), co-moderator/note-taker (Bodurtha or Teague), and equipment assistant (Elena), arrive ~ 45 minutes before each focus group session to set-up the recorder and review the session plan and questions.
- Hold sessions on a weeknight at ~ 7pm.
- Limit sessions to 90 minutes.
- Serve refreshments

***Focus group session content:***

- Upon arrival, have participants complete a consent form and a brief survey containing demographic and three content questions.
- Following the focus group, the staff team has a debriefing session, which includes checking the quality of the tape recording and taking more detailed notes if the recording is of poor quality, recording initial impressions and observations, and completing a debriefing questionnaire.

***Data analysis:***

- Staff reviews the transcripts independently to identify themes, then compares overall impressions and summaries to jointly determine key findings
- A summary of findings for the final GeneSEAN report that discusses key findings and central themes, incorporating direct quotes as examples, is written.

## *FOCUS GROUP INVITATION LETTER*

Dear Sir or Madam:

As a consumer of clinical genetic services, we invite you to be part of a small group discussion regarding the quality of those services. We would also like to hear about your views of genetic healthcare needs in Virginia. Patient feedback is a key part of our project, Virginia *GeneSEAN*. This project involves a broad evaluation of clinical genetic services in the state. Virginia *GeneSEAN* is supported by a grant from the March of Dimes. The ultimate goal is to improve Virginia's genetic services.

A small group session will be held in your local area within the next three months. This is an opportunity to share your views on critical issues affecting families that are impacted by genetic conditions. Your responses will be confidential. Results obtained from the small group activities will not identify individual respondents. All findings will be presented in combined form. If you are interested in participating or would like more information, please contact Elena Zvirbulis. She can be reached at (804) 828-9632, ext.123 (Richmond area) or (800)-___-____ (outside Richmond). We will provide refreshments during the meeting. The meeting will take approximately 90 minutes of your time. Participants will be given a $25 gift certificate following the session as a token of appreciation.

If you have any further questions or would like to participate, please contact us at the phone numbers given above. We look forward to hearing from you!

Sincerely,

Joann Bodurtha, MD, MPH
Principal Investigator

Katherine Teague, MS, CGC
Project Coordinator

Sara Plummer, MSW
Focus Group Moderator

Elena Zvirbulis, BS
Student Assistant

## *PARTICIPANT INQUIRY TELEPHONE SCRIPT*

Hello, I'm Elena Zvirbulis, a graduate student helping with this project. Thanks for calling – let me tell you a little about the project. Then I'll get some information from you.

The purpose of the project, called Virginia GeneSEAN, is to look at what genetic services are available for patients in Virginia. We are also measuring the quality and appropriateness of those services. We hope that our results will guide future planning for genetics in the state. We want to improve genetic healthcare in Virginia – that is the goal.

One very important part of our research involves getting feedback from genetic patients. That is where you come in. We will be having several patient focus groups, or small group discussions, in different parts of the state to get input from individuals who have recently received genetic services. Basically, these focus group sessions will involve about 8-10 patients, plus 2-3 staff involved with the project. The sessions will take approximately 90 minutes. They will include a small amount of time for completing a one page survey and consent forms, then the majority of the time will be spent having a group discussion, led by one of the staff members, about your impressions of the genetic services you have received. The discussion will be tape-recorded. This way we can review participants' responses after the session. No names will be used so no one will be able to identify individual respondents on the tape. No one will be identified in our results, either, since our report will not include names.

The focus group sessions will be held on a weeknight. We will provide refreshments during the session. Participants will also receive a $25 gift certificate after the session as a thank you for participating.

If you're interested in participating, I'd like to get some information from you, but first – do you have any questions for me? *[Answer as appropriate]*

1) Where did you receive genetic services recently? *[Circle answer below]*

| | | |
|---|---|---|
| *Mary Washington Hospital* | → | *Fredericksburg* |
| *Rappahannock Child Development Center* | → | *Fredericksburg* |
| *Virginia Baptist Hospital* | → | *Lynchburg* |
| *Children's Hospital of The King's Daughters* | → | *Norfolk* |
| *Eastern Virginia Medical School* | → | *Norfolk* |

*2)* So you would be interested in our Fredericksburg / Lynchburg / Norfolk focus group session? *[Choose appropriate location based on previous response]*

3) The planned date of that session is:

*Fredericksburg* = _____ beginning at _____PM
*Lynchburg* = _____ beginning at _____PM
*Norfolk* = _____ beginning at _____PM
Are you available then? *[Yes / No / Preference _____]*

4) The meeting will be held at _____

5) Are you 18 or older? *[Yes / No]* *[Note - only adults can participate]*

6) I will be mailing out a confirmation letter with the meeting date, time, location, directions, and parking information. What is your name and mailing address?

   _____

   _____

7) Is there a phone number where I can reach you if necessary? _____ Is it OK to leave a message regarding the focus group session if you are not available when I call? *[Yes / No]*

Thanks very much for your time and interest. Do you have any other questions for me?

***FOCUS GROUP INFORMATION LETTER***

Dear _____,

Thank you for agreeing to participate in the upcoming small group discussion of genetic services in Virginia. We greatly appreciate your willingness to provide feedback. Your input will be used in the development of a report on the status of genetic services and education in Virginia, part of an ongoing project involving a comprehensive assessment of clinical genetic services in the state. The project, Virginia *GeneSEAN,* is supported by a grant from the March of Dimes. Ultimately, the results will aid in the improvement of Virginia's genetic services in the future. We will be tape recording your discussion so we can keep a careful record of the things that we hear from you and the others. Your responses will be confidential and results obtained from the small group activities will not identify individual respondents. All findings will be presented in aggregate form.

Your session will be held at ____ PM on _____, _____, 2004, at _____ in _____. Directions and parking information are enclosed for your convenience. The session will last approximately 90 minutes. We know how valuable your time is, and we will respect everyone's schedules by starting and ending on time. Please allow yourself enough time to get to the meeting on time; if you arrive after the discussion has started, we may not be able to include you. Refreshments will be provided at the meeting and participants will be given a $25 gift certificate following the session as a token of appreciation. Feel free to contact us if you have any further questions at (804) 828-9632 ext.123 (in the Richmond area) or (800) ___-____ (outside Richmond). We look forward to meeting you!

Sincerely,

Joann Bodurtha, MD, MPH
Principal Investigator

Katherine Teague, MS, CGC
Project Coordinator

Sara Plummer, MSW
Focus Group Moderator

Elena Zvirbulis, BS
Student Assistant

## *FOCUS GROUP CONSENT FORM AND SURVEY LETTER*

Thank you for agreeing to participate in this small group discussion of genetic services in Virginia. We greatly appreciate your willingness to provide feedback. Your input will be used in the development of a report on the status of genetic services and education in Virginia, part of an ongoing project involving a comprehensive assessment of clinical genetic services in the state. The project, Virginia *GeneSEAN,* is supported by a grant from the March of Dimes. Ultimately, the results will aid in the improvement of Virginia's genetic services in the future.

Please take a few minutes to complete the enclosed consent form and brief survey and return them to a staff member before the session begins. We will be tape recording your discussion so we can keep a careful record of the things that we hear from you and the others, however, your responses will be confidential. Results obtained from the small group activities will not identify individual respondents. All findings will be presented in aggregate form.

If you have any questions or comments about this project in the future, please feel free to contact us at (804) 828-9632 ext.123. Thank you for your time and interest!

Sincerely,

Joann Bodurtha, MD, MPH
Principal Investigator

Katherine Teague, MS, CGC
Project Coordinator

Sara Plummer, MSW
Focus Group Moderator

Elena Zvirbulis, BS
Student Assistant

**RESEARCH SUBJECT INFORMATION AND CONSENT FORM**

**TITLE:** Virginia GeneSEAN: Genetic Services and Education – Assessment of Needs (Consumer Focus Group)
**VCU IRB NO.:** 3836
**SPONSOR:** March of Dimes, Virginia Chapter

This consent form may contain words that you do not understand. Please ask the study staff to explain any words that you do not clearly understand.

**Purpose of the Study:**
The purpose of this research study is to determine the current status of medical genetic services and education in Virginia. Items of interest include existing systems of care, prevention services, educational activities, insurance issues, data collection, quality assurance and ongoing evaluation, as well as gaps in these systems. You are being asked to participate in this study because you have been identified as a recipient of clinical genetic services in Virginia.

**Description of the Study and Your Involvement:**
If you decide to be in this research study, you will be asked to sign this consent form. First, you should have all of your questions answered and should understand what will happen to you. In this study, you will be asked to 1) complete a brief survey detailing your demographic information and your views about genetic services and 2) participate in a small group discussion about genetic services in Virginia. 7-9 other individuals that have received clinical genetic services in Virginia will also be participating. The meeting will be tape-recorded. However, no names will be recorded on the tape.

**Risks and Discomforts:**
There are no known risks or discomforts involved with participation in this study.

**Benefits:**
There may be no direct benefits to you resulting from participation in this study. However, the resulting data may help improve genetic services and education in Virginia in the future.

**Costs:**
There are no costs for participating in this study other than the time you will spend at the session. It is anticipated that completion of the survey and participation in the small group discussion will take approximately 90 minutes.

**Payment for Participation:**
You will receive a $25 gift certificate at the end of the small group discussion. Refreshments will also be provided during the session.

**Alternatives:**
You have the option not to participate in this study.

**Confidentiality:**
We will not tell anyone the answers you give us. However, other participants will know what you said. The session will be tape-recorded. However, no names will be used. At the beginning of the session, all members will be asked to use initials only so that no names are recorded. The tapes and the notes will be stored in a locked cabinet. After the information from the tapes is typed up, the tapes will be destroyed. The documents and publications developed from the survey and small group discussion data will not identify individual participants.

**Voluntary Participation and Withdrawal:**
Participation in this study is voluntary. If you choose to participate you may stop at any time without any penalty. You may also choose not to answer particular questions that are asked in the survey or small group discussion. Your decision will not change your future medical care at the institutions involved in this study.

**Questions:**
If you have any questions about the study or your participation, contact Dr. Joann Bodurtha at 804-828-9632 or bodurtha@hsc.vcu.edu. If you have any questions about your rights as a participant in this study, you may contact:
Office for Research Subjects Protection
Virginia Commonwealth University
Biotech One
800 East Leigh Street, Suite 114
Richmond, VA 23219
Telephone: 804-828-0868

**Consent:**
I have been given the chance to read this consent form. I understand the information about this study. Questions I wanted to ask about the study have been answered. My signature says that I am willing to participate in this study.

| | | |
|---|---|---|
| _____ | _____ | _____ |
| Participant printed name | Signature | Date |
| _____ | _____ | _____ |
| Witness printed name | Signature | Date |

## *FOCUS GROUP SURVEY*

*Thank you for your participation. Please put a check mark next to the most appropriate answer for each question. Return the completed survey to a staff member before the session begins.*

1) Gender:
____ male
____ female

2) Age:
____ 18-29 years
____ 30-39 years
____ 40-49 years
____ 50-59 years
____ 60 or more years

3) Race/ethnicity:
____ White
____ Black, African American, or Negro
____ American Indian or Alaska Native
____ Asian
____ Native Hawaiian or other Pacific Islander
____ Spanish, Hispanic, or Latino
____ other *(please specify: _____)*

4) Highest level of education or schooling completed:
____ less than high school diploma or the equivalent (GED)
____ high school diploma or the equivalent (GED)
____ some college
____ Associate degree
____ Bachelor's degree
____ Master's degree
____ Doctorate or an equivalent professional degree

5) Please rate the overall quality of clinical genetic services in Virginia.
____ very poor
____ poor
____ neutral
____ good
____ excellent

6) In your opinion, how important is genetics for society?
____ very unimportant
____ somewhat unimportant
____ neutral
____ somewhat important
____ very important

7) How would you rate your knowledge and understanding of genetics?
____ very poor
____ poor
____ neutral
____ good
____ excellent

March of Dimes
Saving babies, together

***Moderator script:***

1) Give a general welcome and thanks. Introduce session staff and invite participants to introduce themselves (initials only). Ask participants to briefly share their connection to genetics, their personal story, if desired.

2) Thanks for sharing… As you know, the main goal of this activity is to obtain consumer input about genetic services in Virginia. To do that, we'd like to get some feedback from you about your overall impressions – positive and negative – of the genetic services you recently received. So think back and describe your opinions of the services.

3) In judging the quality of genetic services, what factors are most important to you?

4) What do you think are the biggest gaps in genetic services in Virginia? What is missing that you think should be available?

5) I'd like to talk about access to care. Could you tell us about any difficulties you have faced in obtaining genetic services? Or were there things that made obtaining care particularly easy?

6) What are your impressions of your primary care providers' and other health care providers' knowledge and awareness of genetic issues?

7) So where do you get information about genetics?

*5 minute break*

8) Solicit input about genetic discrimination and privacy

9) *Do a brainstorm exercise using flip charts.* Let's brainstorm a bit about:
    a) what genetics does well currently
    b) what needs improvement

    *After generating lists* – Now pick the top 3 items in each category.

10) How can genetic services and education in Virginia be improved?

*Have note-taker give a summary of the session and its take-home message(s).*
11) Does that accurately describe your views? Did we miss anything?

12) Finally, do you think this exercise is a useful way to assess genetic services in the state? What might work better?

Thanks very much for your participation!

## *FOCUS GROUP STAFF DEBRIEFING QUESTIONNAIRE*

1. What were the most important themes or ideas discussed?

2. How did these differ from what we expected?

3. How did these differ from what occurred in the previous focus group(s)?

4. Were there any unexpected findings? What surprised us most?

5. What points particularly need to be included in the report?

6. What quotes should be remembered and possibly included in the report?

7. Should we do anything differently for the next focus group?

## Appendix E. Commonwealth Poll Questions

### *GENETIC QUESTIONS FOR COMMONWEALTH POLL*

My first questions are about health issues.

>gene1< Genetics is the study of information, or traits, which are inherited or passed from parent to child. How would you rate your own personal knowledge of genetics on a scale of 1 to 5 where 1 means "I know almost nothing at all" and 5 means "I know everything I need to know"?

<1> I know almost nothing at all
<2>
<3>
<4>
<5> I know everything I need to know
<8> Don't know
<9> No answer

>gene2< Genetic services are offered by professionals to help people in identifying, understanding, and treating inherited conditions. Within the last 5 years, have you received any genetic services (diagnosis of genetic disease, pregnancy screening for birth defects, genetic counseling, genetic testing)?

<1> Yes
<2> No
<8> Don't know
<9> No answer

[#IF NO on gene2 - GO TO gene4]
[#IF DK, NA on gene2 - GO TO gene5]

>gene3< Within the last five years, what kinds of genetic services have you received?

(DO NOT READ. Code into the following categories. If uncertain record verbatim under "Other")

<1> Genetic counseling
<2> Evaluation/diagnosis of a genetic disease or birth defect in a child or adult
<3> Treatment of a genetic disease or birth defect in a child or adult
<4> Screening of a pregnancy for a genetic disease or birth defect
<5> Genetic testing in either a child or an adult
<6> Newborn screening

<66> Other
<88> Don't know (8//)
<99> No answer (9//)

[GO TO gene5]

>gene4< Why have you not used genetic services within the last 5 years?

(DO NOT READ. Code into the following categories. If uncertain record verbatim under "Other")

<1> Services not needed
<2> Was not interested in receiving services
<3> Used genetic services 6 or more years ago
<4> Did not know that services were available
<5> Did not know where to get services
<6> Insurance did not cover services
<7> Could not afford the services
<8> Would have to travel too far to get services

<66> Other
<88> Don't know (8//)
<99> No answer (9//)

>gene5< Have you ever needed genetic information or had questions about genetics?

<1> Yes
<2> No
<8> Don't know
<9> No answer

[#IF NO, DK, NA on gene5 - GO TO gene7]

>gene6< What sources of information have you ever used to get answers to your questions about genetics?

(DO NOT READ. Code into the following categories. If uncertain record verbatim under "Other")

<1> geneticist / genetic counselor
<2> family physician/primary care physician
<3> OB/GYN
<4> other medical specialist
<5> textbook / reference book
<6> Internet
<7> friends
<8> family
<9> health-related TV or radio program
<10> health magazine
<11> college/university faculty
<12> nurse
<13> community lecture
<14> past experience

<66> Other
<88> Don't know (8//)

<99> No answer (9//)

>gene7< If you ever had questions about genetics in the future, what sources of information would you use to get answers to your questions about genetics?

(DO NOT READ. Code into the following categories. If uncertain record verbatim under "Other")

<1> geneticist / genetic counselor
<2> family physician/primary care physician
<3> OB/GYN
<4> other medical specialist
<5> textbook / reference book
<6> Internet
<7> friends
<8> family
<9> health-related TV or radio program
<10> health magazine
<11> college/university faculty
<12> nurse
<13> community lecture
<14> past experience

<66> Other
<88> Don't know (8//)
<99> No answer (9//)

>gene8< In general, how concerned are you about your ability to keep your genetic information private— are you very concerned, somewhat concerned, not too concerned, or not at all concerned?

<1> very concerned
<2> somewhat concerned
<3> not too concerned
<4> not at all concerned
<8> Don't know
<9> No answer

>gene9< What is the maximum number of miles you would be willing to travel to get services from a genetics center?

(DO NOT READ. Code into the following categories.)

<1> Do not want services
<2> 0 miles—would not be willing to travel at all
<3> Less than 5 miles
<4> 5 to 10 miles
<5> 11 to 20 miles
<6> 21 to 30 miles
<7> 31 to 40 miles
<8> 41 to 50 miles
<9> 51 to 75 miles
<10> 76 to 100 miles

<11> more than 100 miles

<88> Don't know (8//)
<99> No answer (9//)

>gene10< Do you know the location of the genetic center nearest to your home?

<1> Yes
<2> No
<9> No answer

>gene11< In your opinion, how important is it to have genetic services available to the citizens of Virginia--is it very important, somewhat important, not very important, or not at all important?

<1> very important
<2> somewhat important
<3> not too important
<4> not at all important

<8> Don't know
<9> No answer

>gene12< Do you have any suggestions for genetic services you would like to see offered in Virginia that currently are not offered?

<1> Yes [GO TO gene13]
<2> No
<8> Don't know
<9> No answer

>gene13< What services would you like to see offered in Virginia?

(RECORD RESPONSE VERBATIM)

8// - Don't know
9// - No answer

## Appendix F. Key Stakeholders Survey

### *STAKEHOLDER LETTER*

September 21, 2005

Dear Stakeholder:

You have been identified as a leader who is shaping the future of Virginia. Your input is needed to help guide healthcare in the Commonwealth. We ask for your help in completing the attached survey on the status of genetic services in our state. This survey is a part of the Virginia GeneSEAN study: Genetic Services and Education, Assessment of Needs. The study is supported by a grant from the March of Dimes. We hope you will help us better understand what is going well and where there are gaps.

For purposes of this study, "genetics" is defined as the study of information, or traits, which are inherited or passed from parent to child. "Genetic providers" are individuals with specialized training in the study of genetics and inherited conditions. The services offered by these professionals to aid families and individuals in identifying, understanding, and treating inherited conditions are known as "genetic services".

Your response to the enclosed survey is important. This is an opportunity to share your views on critical issues affecting families with genetic conditions and genetic susceptibilities. Your responses will be confidential and documents developed from the survey data will not identify individual respondents. All findings will be presented in aggregate form. The ultimate goal of this project is to provide a document which is useful to health professionals, educators, and policymakers engaging in initiatives to improve the quality of genetic health care in Virginia.

Genetics and family history information play an important part in the health of all individuals. However, only one-third of Americans have ever tried to gather their family history. The U.S. Surgeon General (www.surgeongeneral.gov) has begun a Family History Initiative to help stress that knowing a family's medical history can save a life. Attached, please find a family history tool that you can use to help further awareness of genetics and family history.

We greatly appreciate your willingness to complete this survey. Please fax or mail the completed survey to Karen Meister, MS. If you have questions or comments, please feel free to contact us. Thank you for your time and interest.

Sincerely,

Karen Meister, MS
Department of Human Genetics
Virginia Commonwealth University
P.O. Box 980033
Richmond, VA 23298
Phone: (804) 897-6904
Fax: (804) 828-7094
Email: karenmeister72@hotmail.com

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Email: bodurtha@hsc.vcu.edu

March of Dimes
Saving babies, together

## *Key Stakeholder Survey*

*Thank you for participating in our survey. Your responses are valued and important. The survey should take approximately 10 minutes to complete. Please indicate the most appropriate answer for each question.*

1) How would you rate your knowledge of genetics?
- ____ very poor
- ____ poor
- ____ neutral
- ____ good
- ____ excellent

2) How would you rate the average Virginian's knowledge of genetics?
- ____ very poor
- ____ poor
- ____ neutral
- ____ good
- ____ excellent

3) Do you feel that the average Virginian would know where to go to have genetic-based questions answered?
- ____ yes
- ____ no

4) How important is it to have genetic services available to the citizens of Virginia?
- ____ very important
- ____ somewhat important
- ____ neutral
- ____ somewhat unimportant
- ____ very unimportant

5) Are there problems with the accessibility or availability of genetic services in Virginia?
- ____ Yes
- ____ No
- ____ Don't know

6) Is the number of genetic providers practicing in Virginia sufficient to meet the need?
- ____ Yes
- ____ No
- ____ Don't know

7) Is the geographic distribution of genetic providers in Virginia sufficient to meet the need?
- ____ Yes
- ____ No
- ____ Don't know

8) Are genetic services adequately covered by most health insurance plans?
- ____ Yes
- ____ No
- ____ Don't know

9) Are you satisfied with the level of protection current genetic discrimination and privacy laws provide for Virginians?
- ____ Satisfied
- ____ Unsatisfied
- ____ Don't know

10) Please rank the top 3 genetic needs for Virginians:
- ____ more genetic education
- ____ more application of genetic research to clinical problems
- ____ improve coordination of services
- ____ telemedicine capabilities for genetic consultation
- ____ more educational materials for families
- ____ more public discussion of ethical/social/legal issues
- ____ more genetic research
- ____ more genetic providers
- ____ services closer to families
- ____ more family involvement
- ____ more newborn screening tests
- ____ other

11) The prevention of birth defects is one important goal of genetic services. In order to reduce the risk of neural tube defects a woman should be taking 0.4 mg of folic acid daily if she:
- ____ is planning a pregnancy
- ____ is capable of becoming pregnant
- ____ may become pregnant in the future
- ____ all of the above

12) Other issues you wish to raise regarding genetic services in Virginia:

## *RESEARCH SUBJECT INFORMATION AND CONSENT FORM*

**TITLE:** 2005 Virginia GeneSEAN: Genetic Services and Education – Assessment of Needs (Key Stakeholder Survey)

**VCU IRB NO.:** 3291

**SPONSOR:** March of Dimes, Virginia Chapter

**Purpose of the Study:**
The purpose of this research study is to determine the current status of medical genetic services and education in Virginia with regard to existing systems of care, prevention services, educational activities, insurance issues, data collection, quality assurance and ongoing evaluation, as well as gaps in these systems. You are being asked to participate in this study because you have been identified as a leader in the Commonwealth of Virginia.

**Description of the Study and Your Involvement:**
If you decide to be in this research study, complete the enclosed survey only after you understand what is involved and have had all of your questions answered. In this study you will be asked to complete a survey detailing your views on critical issues affecting clinical genetic services and education.

**Risks and Discomforts:**
There are no known risks or discomforts involved with participation in this study.

**Benefits:**
There may be no direct benefits to you resulting from participation in this study, however, the resulting data may help improve genetic services and education in Virginia in the future.

**Costs:**
It is anticipated that completion of the survey will take approximately 10 minutes of your time.

**Alternatives:**
You have the option not to participate in this study.

**Confidentiality:**
Your responses will be confidential and the documents and publications developed from the survey will not identify individual respondents. All findings will be presented in aggregate form.

**Voluntary Participation and Withdrawal:**
Participation in this study is voluntary. You may also choose not to answer particular questions that are asked in the survey.

**Questions:**
If you have any questions about the study or your participation, contact Dr. Joann Bodurtha at 804-828-9632 or bodurtha@hsc.vcu.edu. If you have any questions about your rights as a participant in this study, you may contact:
Office for Research Subjects Protection
Virginia Commonwealth University
1101 E. Marshall St., Room 1-023
P.O. Box 980568
Richmond, VA 23298
Telephone: 804-828-0868

**Consent:**
If you understand this consent, have no questions about the study and agree to participate, no signature is required. Your completion and submission of the survey will serve as your consent. If you agree to participate, please complete and return the survey.

## Virginia GeneSEAN: Genetic Services and Education – An Assessment of Needs

To help assure that we have provided the most comprehensive assessment of medical genetic services and education throughout Virginia, we would appreciate your comments on this report. Please take a few minutes to complete this form and return it to us. Thank you for your feedback.

Return Form to: Joann Bodurtha, MD, MPH
Department of Human Genetics
Virginia Commonwealth University
P.O. Box 980033
Richmond, VA 23298
Fax: (804) 828-7094

Please circle the number that best reflects your answer for each of the following statements.

| | 1 | 2 | 3 | 4 |
|---|---|---|---|---|
| 1. This report is easy to read; that is, it is well organized, and the text is easy to understand. | Strongly Disagree | Disagree | Strongly Agree | Agree |
| 2. The table descriptions, figures, and statistical analyses are clear and appropriate. | Strongly Disagree | Disagree | Strongly Agree | Agree |
| 3. This report helped me to understand the current status of medical genetic services and education in Virginia, as well of areas in need. | Strongly Disagree | Disagree | Strongly Agree | Agree |
| 4. This report will be useful to me in my professional or advocacy role. | Strongly Disagree | Disagree | Strongly Agree | Agree |

Please complete the following statements.

5. The following sections of the report are <u>most</u> helpful (list all that apply): _____

_____

6. The following sections of the report are <u>least</u> helpful (list all that apply): _____

_____

7. The following category best describes my role in relation to this report (please circle one):

| | | | | | | |
|---|---|---|---|---|---|---|
| Genetic Healthcare Provider | Non-Genetic Healthcare Provider | Policy Maker | Insurance Company Representative | Researcher | Consumer | Other: _____ |

8. Other comments or suggestions: _____

_____